

SENSE
about SCIENCE



EUROFORGEN
Network of Excellence



MAKING SENSE OF
**FORENSIC
GENETICS**

What can DNA tell
you about a crime?

Published in 2017

ABOUT THIS GUIDE

Working in Partnership

Sense about Science's public engagement team helps researchers to discuss and present research information, guided by the public and people who will use it. We draw from our extensive public networks and over a decade of working with the public on some of the trickiest issues of evidence. Our ethos is public led, expert fed, which means engaging early and directly addressing what people are thinking. These partnership projects are only available for socially or scientifically difficult issues where researchers make a convincing case that it is a matter of public interest and evidence is neglected, conflicting or misunderstood. In 2016, we were approached by the European Forensic Genetics Network of Excellence (EUROFORGEN-NoE) to help develop their guide to forensic genetics.

A word from EUROFORGEN

This guide is designed to introduce professional and public audiences to the use of DNA in criminal investigations; to understand what DNA can and can't tell us about a crime, and what the current and future uses of DNA analysis in the criminal justice system might be.

Forensic DNA analysis is a complex area open to misinterpretation. So we set out to provide a straightforward guide for the police, judiciary, lawyers, jurors, journalists and those intrigued by criminal casework — in other words, anyone with an interest in the use of DNA for crime investigation. To help us, we approached Sense about Science and formed a public engagement partnership; they connected us with a wide public audience who gave us invaluable feedback on the guide.

Making Sense of Forensic Genetics is the final output of our European Union Seventh Framework Programme funded research and networking project, which has spanned five years and ranged in expertise from forensic geneticists and social scientists to representatives of the judiciary. The European Forensic Genetics Network of Excellence will continue to exist independently from EC funding to provide information and training both to the scientific community and to the interested public.

The contributors' disclosure of interests are available at: senseaboutscience.org/activities/making-sense-of-forensic-genetics

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All our guides are date stamped and reflect the scientific findings and knowledge available at the time of publication.

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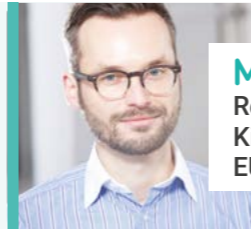
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INTRODUCTION

DNA is present in most cells of our body. It is unique to each of us, and we leave a trail of it everywhere we go. Forensic investigators take advantage of this, using our DNA to draw conclusions about where we've been and who we've interacted with.

In popular television dramas such as *The Killing* and *Midsomer Murders*, the science of forensic **DNA profiling** often helps identify suspects when other lines of evidence have gone cold. Of course DNA analysis has revolutionised forensic science in real life too; helping to catch prolific murderers such as the Green River Killer (see page 15); enabling the remains of those killed by mass disasters and atrocities such as the Srebrenica Massacre to be repatriated to their loved ones; and shining a light on miscarriages of justice that have seen innocent people wrongfully convicted of serious crimes.

Such is the power of DNA to identify, convict, and exonerate, that many perceive it to be infallible. Yet DNA evidence has a number of limitations: it might be undetectable, overlooked, or found in such minute traces as to make interpretation difficult. Its analysis is subject to error and bias. Additionally, **DNA profiles** can be misinterpreted, and their importance exaggerated, as illustrated by the wrongful arrest of a British man, Adam Scott (see page 7). Even if DNA is detected at a crime scene, this doesn't establish guilt. Accordingly, DNA needs to be viewed within a framework of other evidence, rather than as a standalone answer to solving crimes¹.

Forensic scientists take great care to minimise errors, by ensuring their methods have been thoroughly tested, and that they are performed by competent people using properly calibrated equipment and following well-controlled procedures to prevent contamination. Even so, mistakes can happen.

The purpose of this guide is to inform readers about what's currently possible with DNA testing in forensic applications, what its limitations are, and what might be possible in future. It will explain how DNA profiles are generated, what they're used for, and how they can be misconstrued. It will also describe cases where DNA has been a game changer, and turned an investigation around. We hope it will be a useful resource to anyone who works with, or crosses paths with DNA evidence in the criminal justice system.

¹ Wienroth M, Morling N, Williams R (2014) Technological Innovations in Forensic Genetics; Social, Legal and Ethical Aspects. *Recent Advances in DNA and Gene Sequences* 8, 98-103

In October 2011, Adam Scott was arrested and charged with raping a woman in Manchester, UK.

Swabs of the woman's genitals revealed traces of sperm, and one of these swabs yielded a **DNA profile** that matched Mr Scott's. This was the only evidence against him. The forensic scientist who processed the sample said: "It is estimated that the chance of obtaining matching DNA components if the DNA came from someone else unrelated to Adam Scott is approximately 1 in a billion"². But Mr Scott claimed he was in his home town of Plymouth, UK (more than 200 miles away) at the time of the attack, and had never been to Manchester in his life.

When challenged, the scientist claimed the DNA evidence provided: "strong scientific support for the view that Adam Scott had sexual intercourse with [the victim] rather than he did not." However, this was an error. By itself, a DNA profile can't provide any information about the body fluid it came from, or lead to the inference that sexual intercourse took place. Two months after his arrest, mobile phone records came to light that corroborated Mr Scott's version of events; revealing that his mobile phone had been used in Plymouth a few hours after the reported rape. Finally, after five months in custody, he was released.

A subsequent investigation revealed that Mr Scott had become implicated as a result of accidental contamination of samples within the lab. The day before processing samples from the alleged rape victim, the lab had handled a DNA sample from Mr Scott, following a 'spitting incident' in Exeter, UK. Unfortunately the disposable plastic plate used to analyse this sample had been inadvertently reused in the rape case, resulting in the misidentification. The true perpetrator was never found.

This incident highlights two important points for courts:

- a) **DNA should not be used as the sole evidence in a criminal case**^{3,4}
- b) **There is a considerable danger if the importance of the DNA evidence is inappropriately afforded greater weight than other evidence**⁵.



The Adam Scott case is a good example of **confirmation bias** — where inconvenient information to the prosecution is ignored or dismissed. The scientist assumed that because sperm was recovered, *all* of the male DNA must have come from the sperm (when in fact Mr Scott's DNA was a spit sample).

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² Report into the circumstances of a complaint received from the Greater Manchester Police on 7 March 2012 regarding DNA evidence provided by LGC Forensics https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/118941/dna-contam-report.pdf

³ Roth A (2010) Safety in Numbers - Deciding When DNA Alone is Enough to Convict. *New York University Law Review* 85: 1130

⁴ The Crown Prosecution Service (2014) *Guidance on Expert Evidence*, p14

⁵ Gill P (2014) *Misleading DNA evidence: Reasons for Miscarriages of Justice*. 1st ed. Academic Press.

CONTENTS

01 What can we detect? 10

DNA can come from almost all types of biological sources and is analysed using a variety of techniques. Which technique investigators choose depends on the amount of DNA available and the questions they are trying to answer. As forensic DNA techniques have developed over time, their ability to detect smaller and smaller amounts of DNA has increased. This has brought justice to the perpetrators of unsolved crimes, but it also raises the risk of wrongful acquittals and convictions if appropriate safeguards are not in place.

02 Where can we detect DNA? 16

Our DNA is everywhere. We're constantly shedding it, passing it to other people, and moving it around. This means that sometimes DNA detected at a crime scene has nothing to do with the crime. Because of this, investigators need to consider when and how DNA might have been deposited onto a surface or object.

03 Context is key 19

DNA doesn't solve crimes in isolation. DNA profiling is an effective investigative tool to be used within the wider context of all other evidence in a case.

04 What are DNA databases for? 23

Matching DNA profiles from crime scene material with those stored in DNA databases has been one of the most significant innovations in crime fighting in recent history, providing vital intelligence and saving police forces time and money. However, the use of DNA databases has also raised concerns about privacy, data security, and fairness.

05 The meaning of a match 27

Not all DNA matches are equally informative. Just because DNA from a crime scene matches a suspect's DNA, this doesn't necessarily mean they contributed it. Crime stain DNA is often missing some of the markers needed to generate a full DNA profile; in such cases several people may be a 'match', but none may be the contributor. For this reason forensic scientists often employ statistics to convey the meaning of the strength of the evidence.

06 Predicting appearance and biogeographic ancestry from DNA 30

The latest advances in forensic genetics enable externally visible characteristics such as hair or eye colour to be predicted from someone's DNA. This could be a powerful investigative tool, but the possibilities of what is currently achievable have sometimes been exaggerated.

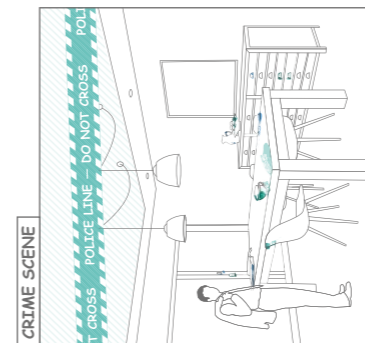
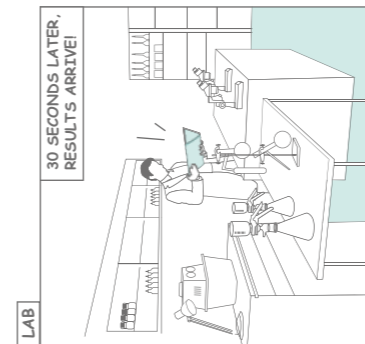
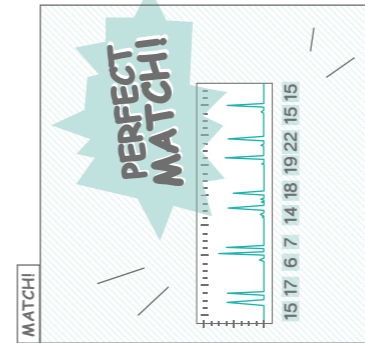
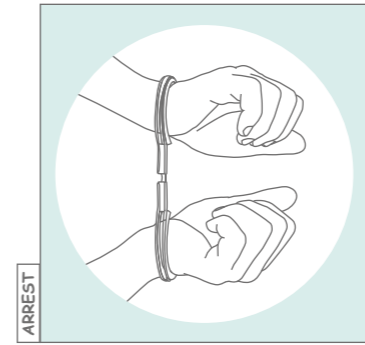
07 Delving deeper 39

More information and sources.

If you see a term that's **green and bold** it's defined in our [Useful Terms to Know](#) on page 37

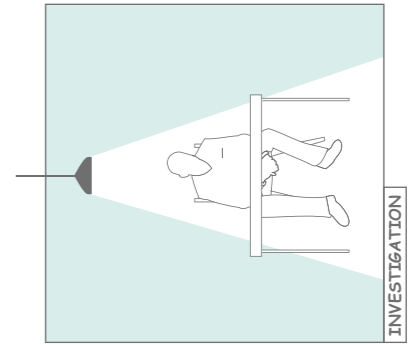
If you see a term that's **blue and bold** it's defined in our [Techniques](#) on page 12

CRIME FICTION vs REALITY

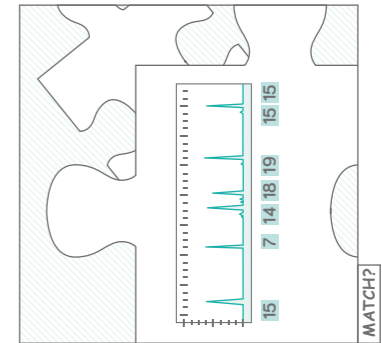


CRIME FICTION

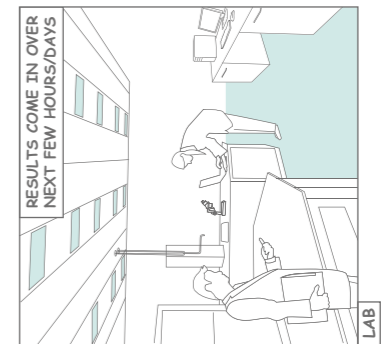
DNA isn't everything, it's only one part of the puzzle



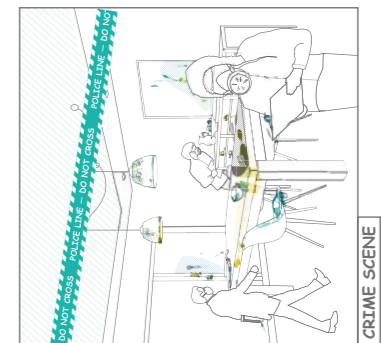
Samples aren't always perfect and we don't always get a match



Results aren't instantaneous and resources aren't infinite



One person does not do everything!



REALITY

01 WHAT CAN WE DETECT?

DNA is a molecule that contains genetic instructions.

It is a major factor in determining the way we look — although 99.9% of our **DNA** is identical to that of other humans. It is the remaining 0.1% that marks us out as individuals, and is therefore of primary interest to forensic geneticists. They can use it to generate a **DNA profile** from human biological material at a crime scene. This can be compared with reference DNA from a named suspect, and a probability that the suspect contributed it can be calculated.

The same DNA sequence is present in every cell of your body (apart from red blood cells), and because you're constantly shedding cells into your environment, this means you leave a trail of DNA behind you. DNA is present in your house dust; in the residue you leave on a glass; and in the root of the hairs stuck to your jumper. Everywhere you go, and everything you touch could contain traces of your DNA.

Until around 2000, forensic geneticists would not have been able to generate a DNA profile from such tiny samples of biological material. But as forensic DNA techniques have developed over time, their sensitivity — or their ability to detect smaller and smaller amounts of DNA — has increased. In the early days, you would have needed a reasonably fresh sample of blood or semen about the size of a British 5 pence piece or European 1 cent coin to generate a DNA profile⁶; today a profile can be generated from just 50 picograms of DNA (the amount contained in roughly 8 human cells). Such traces are invisible to the naked eye.

DNA can be detected and analysed using a number of different forensic techniques, each of which target different parts of DNA. Some, such as **STR profiling** (the most common sort of **DNA profiling** — see the techniques table), target the **nuclear DNA** in our **chromosomes**; others target the small circles of DNA found in cellular energy factories called mitochondria.

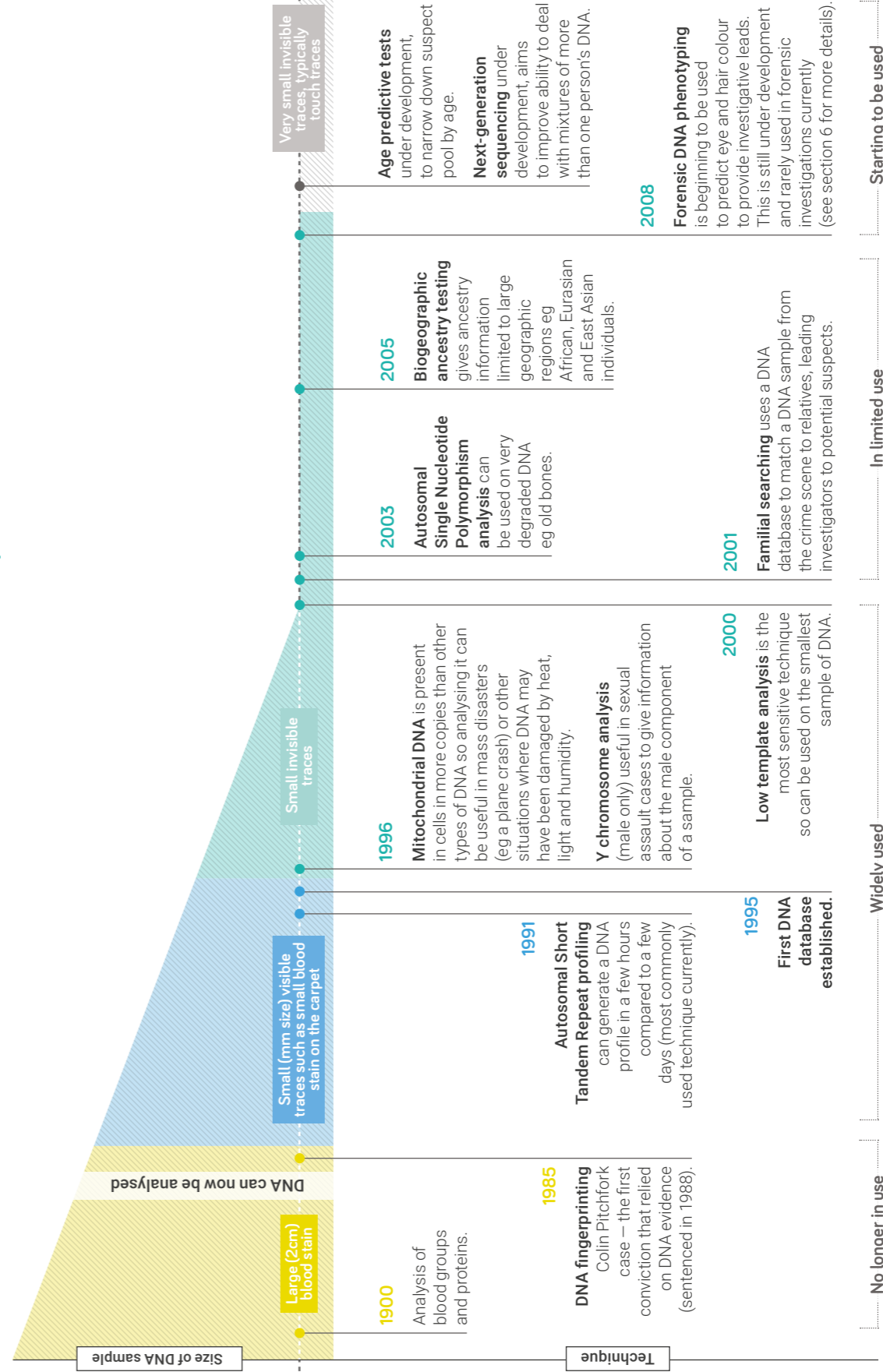
The ease with which DNA profiles can be extracted from different body tissues also varies. It is relatively easy to generate a DNA profile from blood, saliva and semen, but extracting DNA from touched objects when often only a small number of skin cells are present is more challenging. Full STR profiles can be generated from hairs, but only if they contain a root (which has intact cells attached to it). If no root is present — as is often the case with hairs recovered from crime scenes — generating a full STR profile can be difficult, but you might still be able to generate a mitochondrial DNA profile. **Mitochondrial DNA** is less efficient at identifying individuals, but can still provide very useful evidence eg in the identification of burned or badly decomposed human remains. It can also provide useful evidence to eliminate someone from an inquiry if a mismatch is found.

⁶ Silverman M, Thompson T (2014) *Written in blood*. 1st ed. Bantam Press, p297

See diagram p11

WHAT CAN WE DETECT?

DEVELOPMENT OF FORENSIC DNA TECHNIQUES THROUGH TIME...



DNA profiling

The method by which patterns within an individual's DNA can be used to create a **DNA profile**. This is often visualised as a graph with peaks (see page 14). The DNA profile from a crime scene is compared with one from a suspect, and the strength of evidence supporting the identification can be calculated. DNA profiling methods vary according to the type and quantity of DNA available, and the question investigators are trying to answer.

STR profiling

The most commonly applied method of **DNA profiling** that makes use of highly repetitive regions of DNA called **short tandem repeats** found throughout human DNA.

Y chromosome analysis

Makes use of genetic information on the Y chromosome (which only males have). It is especially useful in cases of sexual assault where male and female DNA are mixed, as only the male pattern will show up during analysis. The Y chromosome is inherited from father to son only, which means that all male relatives on the paternal side of the family will normally share the same Y chromosome.

Mitochondrial DNA analysis

Uses DNA from tiny energy factories found inside all human cells called mitochondria. **Mitochondrial DNA (mtDNA)** is more abundant than other types of DNA, and can be useful in cases where biological material is limited (eg if cells have been damaged by environmental exposure, such as heat, light or water, which break up the DNA strand). MtDNA is inherited by a child from its mother, so all relatives on the maternal line of the family will share the same mtDNA.

SNP analysis

Involves the detection of tiny variations in DNA called **single nucleotide polymorphisms**. Because **SNPs** are smaller and more abundant in each cell than **STRs**, SNP analysis can be useful when DNA is highly degraded.

Familial searching

Involves searching a DNA database for profiles that partly match the crime scene **DNA profile**. If a database profile matches to more markers than is expected by chance, it may belong to a relative of the suspect. This technique can therefore generate leads when a full match can't be found.

Low template DNA analysis

A number of techniques (eg low copy number analysis) used to produce a **DNA profile** from very small quantities of crime scene DNA. The standard **STR profiling** technique is modified to enable forensic scientists to look at more of the DNA present.

Biogeographic ancestry testing

A technique that enables an individual's broad geographic origins (eg Africa, Western Eurasia, East Asia, South Asia) to be estimated based on genetic differences in their DNA. This method uses DNA markers that are more or less common in different parts of the world, and can help narrow down a pool of suspects when no match in a **national DNA database** has been found.

Forensic DNA phenotyping

Uses DNA to make predictions about someone's appearance (eg hair colour, eye colour). It is another way of narrowing down a pool of suspects when no match in a **national DNA database** has been found. This technique uses DNA markers found in the genes that determine aspects of human appearance. As it is a new technique, it has so far only been used in a very small number of cases.

Next-generation sequencing

Describes a suite of emerging DNA sequencing technologies, where sensitive tests can be done simultaneously — i.e. you can do **STR profiling**, **biogeographic ancestry testing** and phenotyping tests at the same time. Their use in forensic science is still in its infancy, but in future they should enable more information to be obtained from forensic DNA samples, eg making it easier to distinguish between individual contributors in a mixed sample of DNA.

Because so much of our DNA is virtually identical, forensic scientists don't analyse all of it (this would also be very expensive). Instead, they usually concentrate on short, highly variable regions of repetitive DNA called short tandem repeats or **STRs**. These differ in length between individuals and can be used as **genetic markers** to generate a **DNA profile** that is extremely rare in a population of unrelated individuals).

Typically, markers are examined at a minimum of 16 locations plus a sex marker, or **loci**, in an individual's DNA. These are visualised as a series of peaks on a graph, the position of which corresponds to the length of the STR, and is recorded as a number. Each location has two STRs (because we inherit one version from our mother and one from our father), which means that an individual's genetic profile can be represented as a series of digits, eg 12/13, 13/15, 9/9, 5/8 — a bit like a lottery number. Each pair of digits always corresponds to a specific location on a **chromosome**.

Once a DNA profile has been generated from a crime scene sample, it can then be compared to other profiles, such as DNA from other crime scenes (eg taken with a swab from a biological trace), the suspect's DNA (eg taken at the police station with a swab), the victim's DNA, or to DNA profiles within a **national DNA database**. If the two profiles are identical, this is called a full match; if parts of the profiles match, this is a partial match. Once a match has been declared, the strength of evidence supporting the identification of a named individual can be calculated.

So what is the chance that your DNA will match that of someone else? It depends on how many locations in the DNA (**loci**) you look at. If a forensic scientist looked at just one locus, the probability of this matching the same marker in another individual would be relatively high (between 1 in 20 and 1 in 100).

But as more loci are examined, the probability of two individuals having an exact match decreases rapidly. It's similar to entering a lottery. Many people who buy a ticket will match one number, but the chances of matching all of them and actually winning the lottery is very, very small.

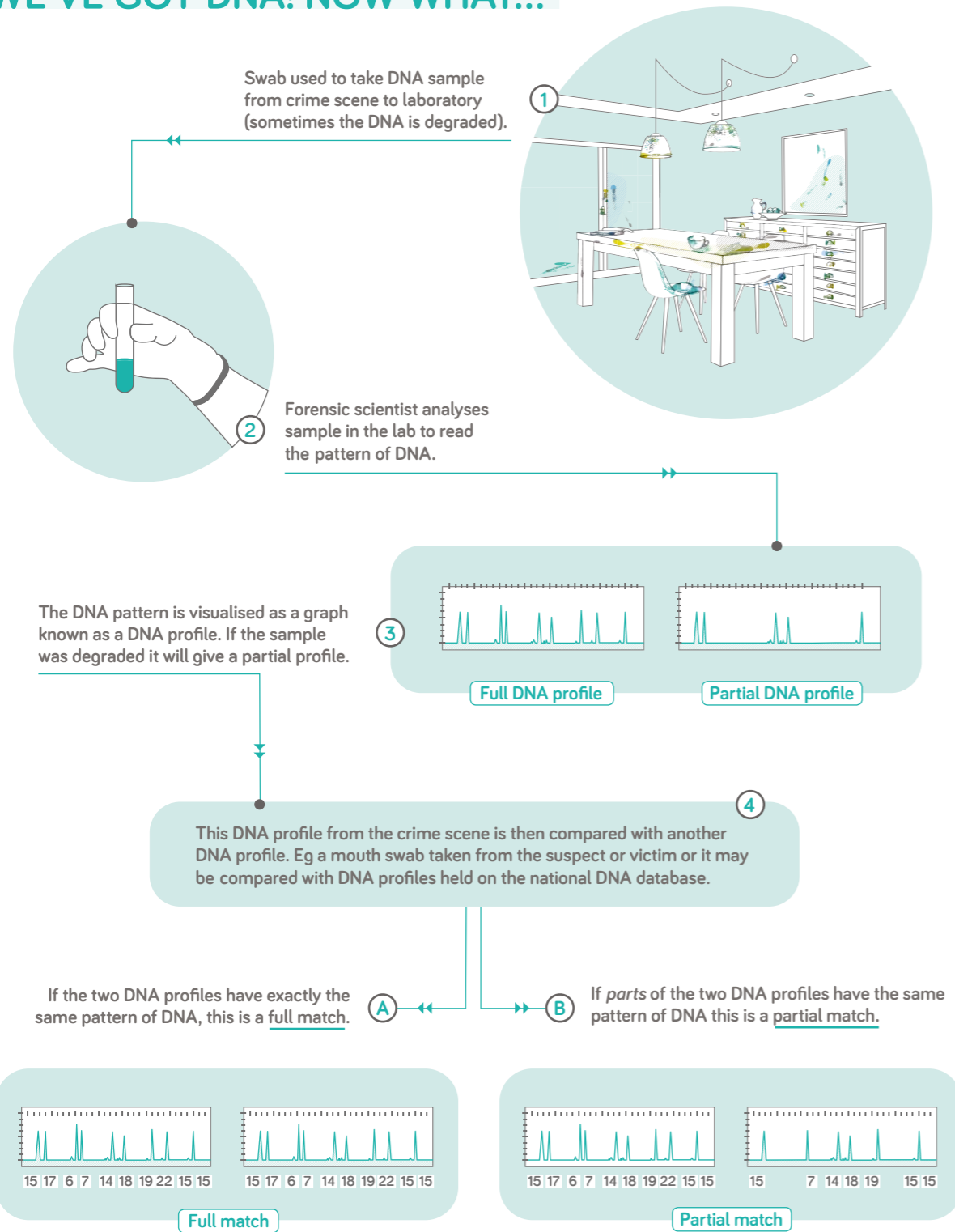
Since European police forces today typically analyse **STRs** at 16 or more loci, the probability that two full DNA profiles match by chance is minuscule — in the region of 1 in 10 with 16 zeros after it (or 1 in 100 million billion)⁷. This means **DNA profiling** can be a very powerful tool in forensic investigations. Although in the UK court, the statistics are always capped at 1 in a billion⁸.

See diagram p14

⁷ Welch LA, Gill P, Phillips C, Ansell R, Morling N, Parson W, Palo JU, Bastisch I (2012) European Network of Forensic Science Institutes (ENFSI): Evaluation of new commercial STR multiplexes that include the European Standard Set (ESS) of markers. *Forensic Science International: Genetics* 6(6): 819-26

⁸ Evett I, Pope S, Puch-Solis R (2016) Providing scientific guidance on DNA to the judiciary. *Science and Justice* 56: 278-81

WE'VE GOT DNA! NOW WHAT...



N.B. This representation of a DNA profile only show 5 pairs of numbers/peaks (genetic markers), but full DNA profiles would actually show at least 16.

The Green River Killings

The development of forensic genetics has been a game changer for certain criminal investigations, including the hunt for one of the most prolific serial killers of all time. Throughout the Eighties and Nineties, the bodies of numerous girls and women were found dumped in overgrown and forested areas near the Green River in Washington State, USA. All had been raped and strangled, but despite the presence of chewing gum and cigarette butts at many of the dump sites, and even traces of semen on some of the victims, it wasn't until 2003 that their killer, Gary Ridgway, was finally caught.

Mr Ridgway first came under suspicion for the killings twenty years earlier, but police had no physical evidence against him. In 1987, with more bodies appearing, police took hair and saliva samples from Mr Ridgway, but still couldn't directly connect him to the victims. It wasn't until 2003 that more sensitive forensic DNA tests finally matched Mr Ridgway to semen found on his earliest victims — women he killed in 1982 and 1983⁹. No-one knows precisely how many women he killed, but he was convicted of 48 murders, and is now in prison for life.

⁹ <http://forensicoutreach.com/library/how-dna-changed-the-course-of-the-green-river-killer-investigation/>



“The more people that appear to be in the mixture, the less sure you can be about the actual number of contributors. For example, about 40% of mixtures from five people actually look like a three person mixture, and virtually none would show a definite indication of 5 contributors because people share many of the same markers.

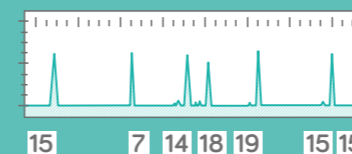
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Partial profiles



Full profile



Partial profile

In an ideal situation you'd have enough DNA to generate a **full DNA profile** (eg using at least 16 loci). However, this isn't always possible. If DNA is only recovered in small amounts, or has been degraded by temperature, moisture, or something else, some **markers** may be missing. This leaves only **partial DNA profiles**. The reduced number of markers makes it more difficult to distinguish between individuals, and the chance of a partial profile matching another DNA profile is much higher.

In addition, the world is a messy place, and DNA is rarely deposited in neat packages from a single person. If a crime scene sample contains the DNA from two or more individuals, then it is referred to as **mixed DNA profile**. Because DNA gets everywhere, all crime scene DNA samples are potential mixtures. This isn't a problem, unless the DNA you're trying to analyse is present at such low levels that it becomes confused with this **background DNA**, or with DNA from another contributor (eg a victim). In such situations, modern computerised methods enable the strength of the evidence to be calculated — something which should be communicated to investigators and juries.

02 WHERE CAN WE DETECT DNA?

DNA gets everywhere. Besides the more obvious methods of DNA transfer, including drops of blood or deposits of semen, small amounts of DNA can also find their way onto people, places and objects via droplets of saliva from talking, sneezing, skin cells shed into house dust or by being left on the surfaces that people touch.

Given how easily DNA can be transferred, this means your DNA could be in a room even if you weren't. If your DNA is found at a location, it could be present because:

- (a) You have been there;
- (b) You touched an object that was later carried to the location by someone else (eg an item of your clothing);
- (c) You encountered a person, who soon after touches something at the location, inadvertently leaving your DNA there (eg you shook hands with them or you both previously touched the same surface).

See diagram p17

As the sensitivity of forensic DNA tests increases, so do the problems...

We've noted that when forensic DNA analysis was first invented in the Eighties, a fairly large sample of biological material was needed to generate a **DNA profile**. But as forensic techniques have improved, their ability to detect smaller and smaller amounts of DNA has increased. This means that tiny, invisible traces of DNA can now be recovered and analysed.

Undoubtedly, this has been beneficial in forensic investigations, enabling difficult cases to be resolved, and the perpetrators brought to justice. However, it is also creating problems. Such is the power of DNA that some crime scene investigators have taken to speculatively swabbing crime scenes, targeting areas they suspect a perpetrator might have had direct contact with, and looking for DNA even when there's no visible stain. This may result in useful investigative leads but it can also result in the detection of DNA that's irrelevant to an investigation, such as a DNA profile from someone who has never visited the crime scene and/or has no connection to the crime. Like any other analytic tool, if used without discrimination, **DNA profiling** can be costly and distracting.



When there's a serious crime, with very little evidence available to indicate the perpetrator, investigators may resort to sampling areas that could have been touched by a potential suspect, such as a door handle or table surface. **DNA profiles** will certainly be found, but it won't be known if any of these belong to the perpetrator, so this could result in false leads.

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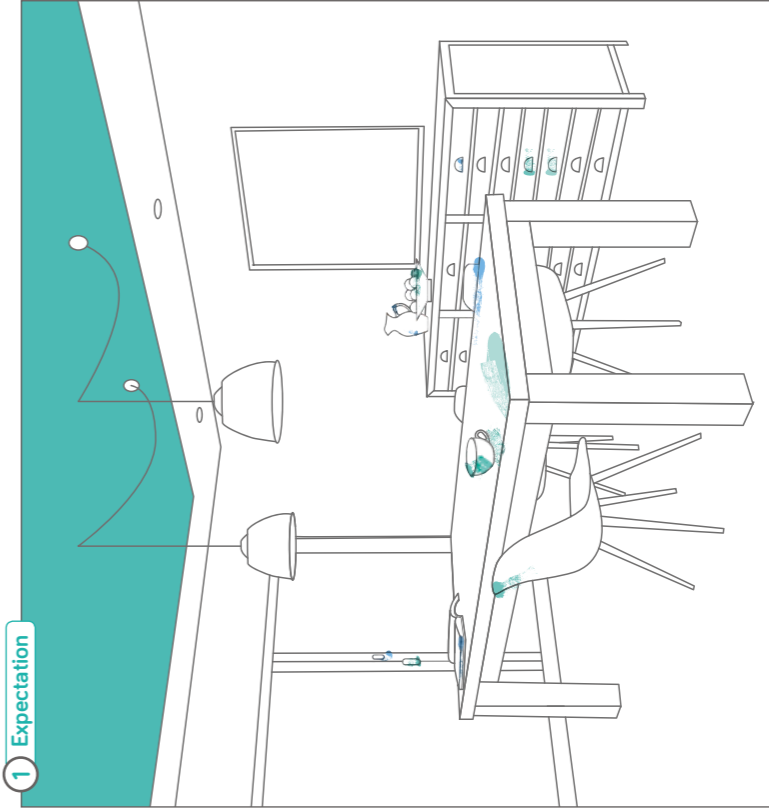
DNA IS EVERYWHERE

DNA is transferred by body fluids, shedding skin cells, talking, coughing and sneezing.

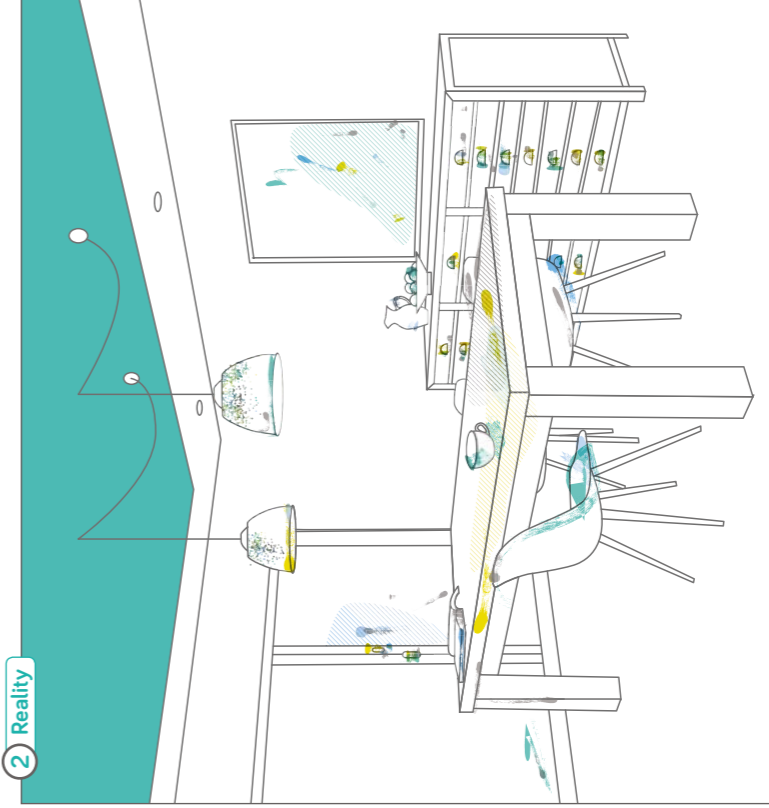
There is even DNA present in house dust.

So DNA from individuals who have nothing to do with a crime might be present at a crime scene. DNA will last almost indefinitely untouched in a dry, cool environment, although activities such as cleaning degrade it. DNA deposited before the crime so unrelated to it is called background DNA.

1 Expectation



2 Reality



The DNA that is present in a room changes rapidly as people and objects interact within it, which is why it's so important for forensic experts to investigate a crime scene as soon as possible so that important DNA does not get moved and new DNA is not introduced.

Each colour represents a different person's DNA

As forensic DNA techniques become more and more sensitive, there's an increased chance that DNA recovered from a **crime scene** is actually;

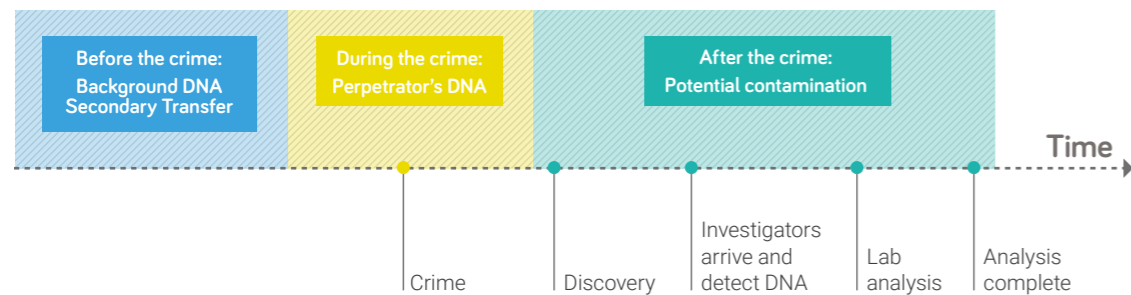
- (a) **Background DNA:** deposited before the crime took place and unrelated to it (see page 17);
- (b) **Secondary transfer DNA:** DNA from someone who was never there, picked up from them by contact and then left at the crime scene by another individual.
- (c) The result of **contamination** by an investigator after the crime took place. Latex gloves can carry DNA and accidentally transfer it between items and locations if an investigator forgets to change them. Other examination tools, such as fingerprinting brushes, can also inadvertently transfer DNA between surfaces. Accidental contamination can also occur within a forensic laboratory (as with the Adam Scott case, see page 7).

Shedder Status

One way in which we release DNA into our environment is through the constant shedding of skin cells onto our clothes and the surfaces we touch. But not everyone does this at the same rate. People who shed lots of skin cells – possibly because of a skin condition such as eczema, dermatitis, dandruff, or even sunburn – are known as 'high status shedders' and are more likely to deposit DNA. For instance, a recent study found that people with atopic dermatitis shed four times as much DNA as healthy individuals¹⁰. Conversely, a 'low status shedder' is less likely to deposit DNA. But not everyone does this at the same rate and it will also vary within the same person at different times.

Another important change that has come with advances in forensic DNA techniques is that when invisible samples are used to generate a **DNA profile**, there's usually no information about *which* body tissue it came from, or *when* the DNA was deposited. When larger amounts of DNA were needed to generate a DNA profile, it was usually possible to run other tests to determine whether a fluid was saliva or semen, say. And in the case of blood, its colour could provide clues about how fresh it was. These tests can't be done on really small biological samples, but DNA can often still be detected.

WHEN COULD DNA BE DEPOSITED?



¹⁰ Kamphausen T et al (2012) International Journal of Legal Medicine 126(1): 179-83

The presence of DNA doesn't necessarily tell us when or how it got there. But it can still be a phenomenally useful tool to police investigations. What matters is *context*.

Some types of DNA evidence are less likely to have been deposited through innocent means than others. For example, a visible blood stain is not so easily transferred unnoticed as an invisible smear of saliva, or a smattering of skin cells.

Questions such as: 'When and how was the DNA deposited on to the surfaces tested?' and: 'How was the DNA collected by crime scene investigators?' are crucial to understanding whether DNA is relevant to an investigation; or if it's **background DNA**; the result of **secondary transfer**; or **contamination**. Additional context may also be provided by other, non-DNA evidence, such as fibres, footwear marks, or fingerprints.

Activity and context

If a large, visible, sample of body fluid, such as a blood stain, is found at a crime scene, then it is easy to obtain DNA from it. And if that blood is found on smashed window glass, say, then there is a good chance it is relevant to the investigation. The suspect may have cut himself while breaking into the property, for example. In other words, the evidence is considered relevant because it is directly associated with the *activity* of the crime – in this case breaking a window. Contrast this with a **DNA profile** taken from a surface, such as a kitchen table, where there's no identifiable body fluid. It is more difficult to propose an activity that could explain its presence because of the lack of *context*.

See diagram p20

So... DNA alone doesn't solve crimes. It's an important detection tool, but it's certainly not a detective.



Other types of forensic evidence can provide important corroboration of DNA results, and vice versa. As an example: fibre analysis depends on having a reference sample from the textile source. If fibres have been recovered from a victim not matching the victim's clothes, there is not much that can be done. However, these fibres may suddenly become informative if a suspect has been identified by **DNA profiling**, and clothes are found in the suspect's apartment matching the fibres from the victim. Then you have corroborative evidence.

Peter Schneider Professor of Forensic Molecular Genetics, Institute of Legal Medicine, University of Cologne
EUROFORGEN Consortium member

Background DNA?

Scenario

A man is found dead at his home, and your DNA is recovered from the crime scene. However, you knew the victim, and often visited him, so your DNA could have been there for weeks before he was killed. This is known as **background DNA**.

Real-life example

Meredith Kercher was stabbed to death in Perugia¹¹, Italy, in 2007, and her flat-mate, Amanda Knox, was a key suspect. A knife retrieved from Ms Knox's boyfriend's flat contained small traces of Ms Kercher's DNA on the blade, and a **DNA profile** from Ms Knox was recovered from the handle. Prosecutors suggested that that the DNA was transferred to the knife when Ms Kercher was stabbed with it, although no blood was detected. Possibly, this was because the knife was cleaned with bleach, they argued. This is a classic example of **confirmation bias**, (see Adam Scott case, see page 7), a psychological effect where an individual(s) fits the evidence to a presupposed set of circumstances, while ignoring other possibilities. For instance, Ms Knox could have used the knife to cut bread (starch grains were also observed on the blade), and since she co-habited with Ms Kercher, there was a ready explanation for the presence of her DNA on the knife blade. Methods used to collect and store the evidence were also found to be sub-standard, so the DNA could also have got there through cross-contamination. In the final judgement, the court accepted the defence version of events and exonerated the defendants.

Secondary transfer?

Scenario

A man is attacked by someone wearing a mask, while walking home from a party. Your DNA is found on the victim's hands. You were also at the party, and had no direct contact with the victim, but you both poured a glass of wine from the same bottle. This caused your DNA to be transferred to their hands. This is known as **secondary transfer**.

Real-life example

When local taxi driver David Butler's DNA was recovered from the fingernails of murdered sex worker Anne Marie Foy, it seemed like an open and shut case. It was presumed that Ms Foy had torn at his skin as he hit and strangled her, before dumping her body in a park near Liverpool city centre in September 2005. The amount of DNA found by police was tiny, but enough to generate a hit against the UK's DNA database (see next chapter), identifying Mr Butler as the source. He denied ever having met the victim, but even though other evidence was lacking, the DNA evidence was enough to see him charged with murder.

However, Mr Butler's defence team queried precisely how that DNA got onto the victim's nails. They established that he was sometimes known by the nickname "flaky", because of the dry skin condition he suffered from, and suggested that perhaps some of his skin cells had transferred to bank notes that were later used to pay Ms Foy – an example of secondary DNA transfer – or they had been transferred to her through other innocent means¹². Mr Butler was acquitted.

¹¹ Gill P (2016) Analysis and implications of the miscarriages of justice of Amanda Knox and Raffaele Sollecito. Forensic Science International: Genetics 23: 9-18

¹² <http://www.telegraph.co.uk/news/science/9115916/The-case-against-DNA.html> (accessed January 2017)

WHO ARE WE DISCOVERING AT THE SCENE? HOW DID MY DNA GET THERE?

DNA is everywhere and context is everything. There have been major developments in the techniques for identifying individuals from samples of DNA. Since the 1990s tests to detect invisible DNA have been developed. Yet, this ability to measure smaller and smaller samples has come with problems. With invisible traces it is not always possible to know whether the DNA came from a body fluid, skin or hair. There is also no time stamp on DNA whereas a visible stain eg blood might give some indication of when it appeared. So, as techniques have improved, the importance of evaluating DNA in context of other information and evidence has also increased.



Sample size

A large visible trace; BLOOD STAIN (2cm) was found at the scene.

A large amount of DNA was obtained from the stain (some micrograms).

A DNA profile was generated and a match with your DNA was found!

Context The type of body tissue (blood) is known and a very general idea of how old the blood stain can be estimated (range: hours vs days).
Conclusion It's highly likely you were there at the time, or recently.

Scenario You were at the premises – you cut yourself on broken glass.

A small visible trace; a tiny STAIN (millimetre size) was found at the scene.

A small amount of DNA was obtained (some nanograms) from the stain.

A DNA profile was generated and a match with your DNA was found!

Context Using a screening test it is possible to identify the type of body tissue from the stain eg blood, semen or saliva without 'consuming' the trace. This means some of the DNA remains intact for future tests.

Conclusion Highly likely you had been to the premises where the crime was committed (but not necessarily recently).

Scenario You'd had a nose bleed in the morning, then went to the premises in the afternoon and sneezed. The crime was committed in the evening.

A small invisible trace (could be from body fluids or skin flakes), was found at the scene.

A small amount of DNA was obtained (several hundred picograms) from the trace.

A DNA profile was generated and a match with your DNA was found!

Context The type of body tissue cannot be determined without consuming the trace (using the entire sample) in the test because there are currently no screening tests for invisible traces.

Conclusion Were you there or not?...

Scenario An invisible trace could have been left during the crime, before or after, or you may have never been at the crime scene.

Scenario You had drunk from a glass at the premises, but it could have been days or even months before (background DNA).

A very small invisible trace; a 'touch' trace was found at the scene.

A very small amount of DNA was obtained (less than a hundred picograms) from the trace.

A partial DNA profile was generated and a match with your DNA was found!

Context The type of body tissue cannot be determined without consuming the trace (using the entire sample) in the test because there are currently no screening tests for invisible traces.

Conclusion Were you there or not?...

Scenario An invisible trace could have been left during the crime, before or after or you may have never been at the crime scene.

Scenario The victim touched the same glass as you at a pub before he/she was attacked (secondary transfer).

Importance of context

Scenario

Who? When? What? Where?

Who? When? What? Where?

Who? When? What? Where?

Contamination?

Scenario As a security guard, you had legitimate access to a building, before a stabbing took place there. The police investigator wore plastic gloves to recover a knife from the crime scene, but first touched a door handle that you had also previously touched. As a result, your DNA was accidentally transferred to the knife. This is known as **contamination**.

Real-life example Farah Jama, was wrongfully convicted of raping a woman in Melbourne, Australia, in 2008 and spent 15 months in prison after a sample of his DNA had contaminated a sample taken from the alleged rape victim¹³. The mistake is believed to have occurred because 28 hours earlier, the same forensic medical officer had taken a DNA swab from a woman with whom Mr Jama had had sex (no charges were made)¹⁴. The precise mechanism of contamination is unknown, but as the two samples were not 'mixed up', it is most likely that the examination room or the equipment used were not cleaned.

¹³ Inquiry Into the Circumstances that led to the Conviction of Mr Farah Abdulkadir Jama <http://www.parliament.vic.gov.au/papers/govpub/VPARL2006-10No301.pdf>

¹⁴ <http://www.smh.com.au/national/dna-fiasco-rape-conviction-quashed-20091207-kfc3.html> (accessed January 2017)

04 WHAT ARE DNA DATABASES FOR?

Once a **DNA profile** has been generated from crime scene material, the next step is to compare it to **DNA profiles of known individuals in order to find a match** – eg to a known suspect. Or in many cases, where no suspect has been identified, this will involve searching a **DNA database**.

Most European countries have their own **national DNA database**. The circumstances under which an individual's DNA can be taken, and whose **DNA profiles** can be retained on such databases vary widely between different countries. In many places, samples can be taken on arrest, but DNA profiles can only be retained for a short period of time - unless the person is convicted of a serious crime. In other countries, DNA profiles can be retained from individuals convicted of *any* offence. National DNA databases also hold DNA profiles retrieved from crime scenes, in case someone whose DNA isn't currently on the database is arrested in the future. This also enables investigators to link crimes which may have been committed by the same person.

Although many national DNA databases are large, they don't contain DNA from everyone living in a country. This means that even if DNA is retrieved from a crime scene, unless the perpetrator's DNA is already in the database, it won't necessarily generate a 'hit' – or match.

The UK National DNA Database by Numbers¹⁵

The UK national DNA database (NDNAD) was launched in 1995 and holds **5 million** DNA profiles from individuals, (plus about half a million crime scene profiles) – equivalent to **nine percent** of the UK population – (or 14% of the male population since 80% of profiles are from men)¹⁵.

About **40** vetted home office officials have access to the database. Police forces do not have access, although they own the records on the database and receive notifications of any matches. The database can only be used to prevent and investigate crime, to prosecute those accused and to identify deceased persons¹⁶.

The UK NDNAD generates matches or hits for more than **32,000** crimes per year.

In 2014/15, the chance of a crime scene profile matching a subject on the database was **63.2%**.

Even so, the likelihood of crime scene DNA being retrieved and sent for testing is low, and forensic analysis tends to be reserved for serious cases eg homicides. For example, in 2014/15, crime scene investigators were sent to look for forensic evidence in **96%** of homicide crime scenes, and found DNA at **65%** of those examined, whereas they were only sent to **27%** of vehicle thefts, and just **30%** of these yielded DNA evidence.

¹⁵ National DNA Database Strategy Board Annual Report 2014/15 https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/484937/52921_NPCC_National_DNA_Database_web_pdf.pdf

¹⁶ Office for National Statistics: United Kingdom population mid-year estimate <https://www.ons.gov.uk/peoplepopulationandcommunity/populationandmigration/populationestimates/timeseries/ukpop/pop> (Accessed January 2017)

Social and ethical aspects of DNA databases

There are clear benefits to maintaining a forensic **DNA database** and some people think they should hold information from everyone in a country. But others have concerns about privacy, data security, and fairness. Questions about whose **DNA profiles** should be retained on the DNA database; for how long; for what purposes; and who should have access to, and oversight of national DNA databases, continue to be debated in many countries. These questions grow more pertinent as interest in the sharing of information across national borders — and even across different types of databases (eg medical and commercial information) — increases, because each database has different security controls, access policies and retention periods.

There are also concerns that certain minority groups are disproportionately represented on national DNA databases. Some argue that such inequalities could stoke feelings that certain groups are being unfairly criminalised or discriminated against^{17,18}.

The right to privacy

Should governments have the right to hold the DNA of people without a conviction indefinitely? Some people have argued that if everyone in a country were held on a national DNA database, far more crimes would be solved, but many are opposed to this idea on the grounds of personal privacy and human dignity. Instead, many countries only keep DNA profiles on the national DNA database from people who have been convicted, or who choose to give their DNA voluntarily.

So what if you're arrested, but not guilty of the crime? In 2001, an 11-year old boy, known as S., and another British man called Mr Marper, were arrested for separate offences, but neither was charged. Both had DNA swabs taken from the inside of their cheeks and their samples were added to the UK national DNA database (NDNAD). After their release, they applied to have their DNA profiles (and fingerprints) removed from police databases including the NDNAD, but these applications were rejected by British Appeal Courts.

In 2008, the European Court of Human Rights asserted that the retention of fingerprints, cellular samples and DNA in such circumstances was in breach of Article 8 of the European Convention on Human Rights — the right to respect for privacy and family life¹⁹. As a result, there has been a change to UK law, meaning that people who have been arrested for, but not charged with, imprisonable offences (and certain non-prisonable ones), must normally have their DNA profiles removed from the DNA database within 3 years²⁰.

¹⁷ Duster T (2004) Selective Arrests, an Ever-Expanding DNA Forensic Database and the Specter of an Early-Twenty-First Century Equivalent of Phrenology in DNA and the Criminal Justice System: The Technology of Justice, ed. David Lazer (Cambridge M.A: MIT Press), p315-334

¹⁸ Policy Equality Statement for the National DNA Database www.gov.uk/government/uploads/system/uploads/attachment_data/file/245240/Policy_Equality_Statement_for_the_National_DNA_Database_Sep13.pdf

¹⁹ <https://justice.org.uk/s-marper-v-uk-2008/> (accessed January 2017)

²⁰ Protection of Freedoms Act 2012: how DNA and fingerprint evidence is protected in law <https://www.gov.uk/government/publications/protection-of-freedoms-act-2012-dna-and-fingerprint-provisions/protection-of-freedoms-act-2012-how-dna-and-fingerprint-evidence-is-protected-in-law> (accessed January 2017)

Match vs identity

Suppose a **DNA profile** from a crime scene is compared against a **national DNA database**. If a match occurs, it's either because that person's DNA was found at the crime scene, there was contamination, or it's a false positive match (a chance match with an individual who was not involved in the crime). The next step is to calculate the strength of the DNA evidence to support the proposition that a specified named individual has contributed to the sample.

Although the chances of two full DNA profiles from two different unrelated individuals matching are extremely small, DNA profiles from crime scenes are rarely perfect, and they may not contain information about every **genetic marker** analysed (i.e. they are a **partial DNA profile**). The smaller the number of genetic markers the DNA profile is composed of, the greater the risk of a false match occurring.

In 1999, Raymond Easton, a 49-year-old man from Swindon, UK, was arrested and charged with a burglary in Bolton, UK (~175 miles away), after a DNA sample from the crime scene matched his DNA profile in the UK national DNA database. Mr Easton was in the advanced stages of Parkinson's disease, and was unable to walk more than ten metres without help. His DNA profile had been loaded onto the database four years earlier following a domestic dispute. The crime scene sample matched Mr Easton's DNA profile at six **loci**, which was considered enough to secure an identification at that time — although the total number of **STRs** required has since been extended to 16 plus a sex marker. The chances of a match was reported as 37-million-to-one²¹. Mr Easton spent several months in custody before his solicitor persuaded police to run further DNA tests, which eliminated him²².

Importantly, **a match does not imply identity**, something we discuss further in chapter 5. Even with a probability as low as one-in-37-million²³, you could find one or more innocent British men in the UK population who would match the crime scene profile described above. The larger the database, the higher the risks of false positive matches, which is why it is essential to consider the non-DNA evidence as well. If this is not done there is a danger that the DNA evidence will be over-weighted by a jury.

The Raymond Easton case emphasises the need to regard DNA evidence as *investigative* in the first instance, and not a panacea for a prosecution. This is not to say it can't be valuable or form an important part of a conviction, but the *context* of the DNA evidence and whether it can be corroborated with other non-DNA evidence, such as fibres, fingerprints, or eyewitness statements, matters and needs to be considered if these are available in a case.

²¹ http://www.heraldsotland.com/news/12440889.Guilty_by_a_handshake__Crime_scene_DNA_tests_may_not_be_as_accurate_as_we_are_led_to_believe/

²² Huff C, Killias M (2008) Wrongful conviction. 1st ed. Philadelphia: Temple University Press

²³ Gill P (2014) Misleading DNA evidence: Reasons for Miscarriages of Justice. 1st ed. Academic Press

Familial searching

If investigators can't match a crime scene **DNA profile** to someone on a **national DNA database**, or to a suspect who has been identified through other means, they may occasionally search for relatives of the person who deposited the DNA by looking for similar DNA matches. This is based on the principle that unrelated people have relatively few DNA markers in common, whereas related people will have more – a parent and child, for instance, will match at least half of the **genetic markers** analysed, as they share 50% of their **autosomal DNA**. As we've noted, we have two copies of DNA at each **locus**, because we inherit one copy from each parent. So a parent and child always share one of the two digits (representing an STR repeat of the same length) at each **locus**.

The more distantly related people are, the less similar their DNA is. This means with standard (autosomal) **DNA profiling, familial searching** is only useful for finding close relatives (such as parent/child/sibling). It typically generates many false leads, so investigators will often limit the size of the pool they search within, eg by confining it to a specific geographic area, in order to produce a useful shortlist to follow up²⁴. Investigators can then look at other evidence that might imply a relative of an individual was involved. Familial searching is only used for very serious crimes, and is allowed in some countries including the UK and the Netherlands, but not, for example, Germany. In the UK, there were only 16 familial searches conducted during 2014-15²⁵.

Familial searching raises ethical questions, because it risks casting suspicion on family members who were not involved in the offence. However, it can sometimes provide an investigative lead where none existed before, and has led to successful prosecutions.

Familial searching in action: Finding justice for Lynette White

Lynette White was a 21-year-old prostitute who was stabbed to death on Valentine's Day, 1988, in Cardiff, UK. Three local men, Stephen Miller, Tony Paris and Yusef Abdullahi, were wrongfully convicted of her murder and spent two years in prison, before their conviction was quashed. The case was reopened in 2000, and thanks to newer, more sensitive methods of DNA analysis, a DNA profile could be obtained from blood on a skirting board that had been found near to her body. This was run against the national DNA database, but no full matches were found. However, there was a partial DNA match with a local 14-year-old boy, who was already known to police. He was only 2 years old when Ms White was murdered, but was possibly related to the killer. When police took DNA samples from other members of his family, the boy's uncle, Jeffrey Gafoor, provided a full match. Soon afterwards Mr Gafoor confessed, and was sentenced to life imprisonment²⁶.

²⁴ Familial Searching, inferring ethnicity and research uses <http://nuffieldbioethics.org/wp-content/uploads/Bioinformatics-Chapter-6-Familial-searching-inferring-ethnicity-and-research-uses.pdf>

²⁵ National DNA Database Strategy Board Annual Report 2014/15 https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/484937/52921_NPCC_National_DNA_Database_web_pdf.pdf

²⁶ <http://thejusticegap.com/2011/12/what-price-justice-the-cardiff-3/> (Accessed January 2017)

DNA doesn't give a simple 'yes' or 'no' answer.

Forensic geneticists typically use 16 or more **genetic markers**, selected from the human genome, plus an indicator of sex, to generate an individual's **DNA profile**. These markers have been chosen because they are extremely variable, so if you have a full DNA profile (with information available for all 16 markers) the chances of finding another unrelated person with exactly the same DNA sequence at each location is very small. This means that the risk of DNA retrieved from a crime scene matching someone unrelated to the true source is extremely low (less than 1 in a billion, and often many orders of magnitude lower than this).

However, many of the DNA profiles retrieved from crime scenes aren't full DNA profiles because they're missing some genetic markers or there is a mixture of DNA from two or more people. So *was it* the suspect who left their DNA at the crime scene? The DNA evidence won't give a 'yes' or 'no' answer: it can only ever be expressed in terms of probability.

How certain?

A **DNA profile** is said to 'match' if all of the markers in the crime stain profile are the same as the markers in the sample it's being compared to (the reference sample). Remember that this is a matching of samples, not a matching of a sample to a specific person. It is possible that several individuals in a database may 'match' a crime scene DNA profile, when none of them actually deposited it.

Once a DNA sample has been analysed, two kinds of statistics may be reported. The simplest is the **match probability**, which addresses the question of how rare a DNA profile is in a population of random unrelated individuals. This must not be confused (but often is) with how likely the person is to be innocent of the crime. For example, if a DNA profile from the crime scene matches the suspect's DNA and the probability of such a match is 1 in 100 million if the DNA came from someone else, this does not mean that the chance of the suspect being innocent is 1 in 100 million. This serious misinterpretation is known as the **prosecutor's fallacy**.

These match probabilities are fine if you are dealing with full DNA profiles from a single individual and they can often be used for a **partial DNA profile** too. But this figure cannot be used for mixtures of two or more individuals or if information about some of the **genetic markers** is missing. Under these circumstances a second kind of analysis is applied: a **likelihood ratio**.

A likelihood ratio weighs the evidence in favour of competing 'stories' (or hypotheses), one from the prosecution perspective and one from the defence. It compares how probable the observed evidence is under each story.

So in the case of a DNA profile from a sample of blood, you would be looking at:

- Assuming the blood comes from the suspect, what is the probability of seeing the match?
- Assuming the blood was someone else's, what is the probability of seeing the match?

The likelihood ratio is obtained by dividing (a) by (b). If the answer is larger than one the prosecution's version of events is better supported; if it's lower than one then the defendant's is.

The guidance given alongside is usually:

Ratio	Expert guidance: the forensic findings...
1	... do not support one proposition over the other
2-10	... provide weak support
10-100	... provide moderate support
100-1,000	... provide moderately strong support
1,000-10,000	... provide strong support
10,000-1 million	... provide very strong support
Over 1 million	... provide extremely strong support ²⁷

Although statistical experts agree that likelihood ratios are the best approach for complex DNA profiles, their adoption by many labs has proved slow. In part this is because of fears that courts may misunderstand the ratio.

Remember: DNA doesn't give 'yes' or 'no' answers, but rather enables us to assess probabilities. So DNA evidence can be very strong, very weak and everything in between.

²⁷ ENFSI Guideline for Evaluative Reporting in Forensic Science <http://www.forensic-isotopes.org/assets/ENFSI%20Guideline%20Evaluative%20Reporting%20March%202015.pdf>

Calculating complexity

Because of the complexity of the calculations – particularly once several people's DNA gets mixed together – DNA analysts now use specially designed computer programs. Just like the physical methods of analysing DNA, these programs need to be **validated** to ensure they have been properly formulated and use theory that is widely accepted by the scientific community.

A number of such programs are in use around the world, but because different programs are prepared using different mathematical approaches and assumptions about the data, they can give different values for the **likelihood ratio**. There have been cases where the prosecution has used one program and the defence another, meaning the court has been presented with two different answers about the strength of the DNA evidence. Usually the difference is only small, but cases where one program favours the prosecution and another favours the defence are obviously important and need further investigation.

It is important to note that several computer programs have been developed by commercial companies, and may not be as available for use by defence experts because they have less money to pay for them. This could make it difficult to scrutinise how the prosecution generated its statistics. Since open source software is freely available and is more transparent, there is currently a debate within the forensic community on the relative benefits of commercial vs open source software.

How reliable is forensic DNA profiling?

In recent years, confidence in other (non-DNA) forensic techniques, such as bite mark analysis or hair microscopy, has been damaged by the discovery that different analysts can reach different conclusions about the same piece of evidence. Some of these older forensic techniques were never properly **validated**, or subjected to rigorous scientific examination, before entering the criminal justice system. They involved a lot of subjectivity.

Ideally, analytical techniques used in forensic science should be validated before they're allowed into court. For a method to be validated it must have a scientific backing that is underpinned by peer reviewed papers in the scientific literature. An important part of testing is to show that when a completely unconnected individual is compared with the 'crime stain' **DNA profile**, the **likelihood ratio** obtained is well below 1 and never very high. Any limitations of the test must also be made clear.

The most important feature distinguishing **DNA profiling** from other non-DNA forensic identification techniques is that for DNA profiling we can base calculations on the exceptionally well understood established theory of genetics. No such underpinning theory is available for most non-DNA forensic techniques.

06 PREDICTING APPEARANCE AND BIOGEOGRAPHIC ANCESTRY FROM DNA

Our DNA underpins many aspects of the way we look, but scientists have only recently begun to get to grips with how individual genes produce physical traits such as hair or eye colour.

A practical application of this work is the development of forensic DNA tests that can predict aspects of someone's physical appearance. This approach is called **forensic DNA phenotyping**²⁸.

But not all externally visible characteristics are equally predictable from DNA information — at least not today. The simplest is biological sex, because females carry two X **chromosomes**, and males carry one X and one Y, which is easily detectable by DNA testing. Eye colour is already harder, because it's influenced by many genes, of which six are currently used in forensic DNA phenotyping tests. Predicting other externally visible traits such as height is even harder, and currently not yet possible because they are determined by large numbers of genes, many of which remain unknown. They are also influenced by environmental factors such as nutrition, which cannot be predicted from DNA.

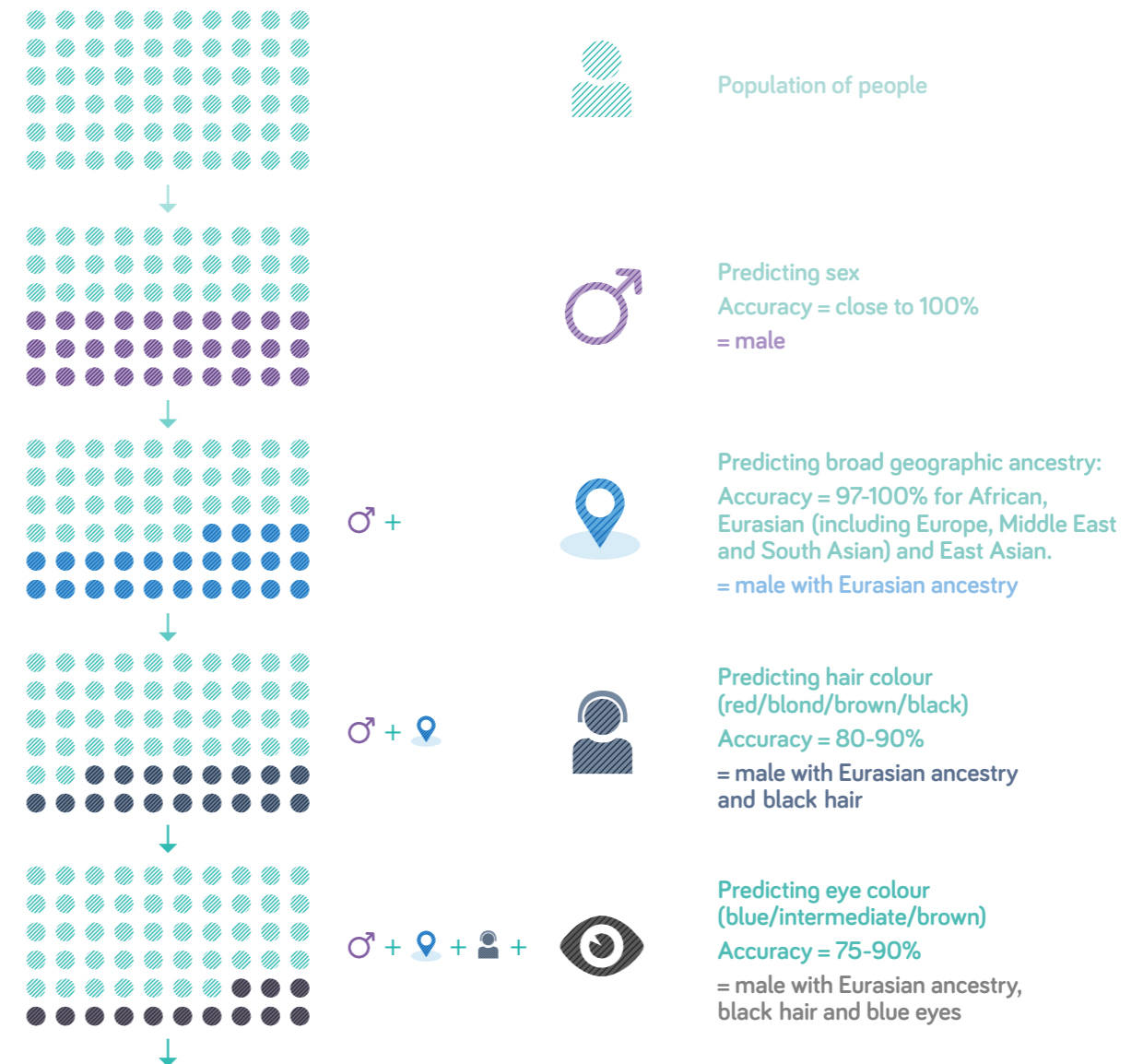
Because human geneticists have only understood the genetic basis of a few externally visible characteristics so far, forensic DNA phenotyping is still in its infancy. In general, it's only used in cases where a **DNA profile** from crime scene material does not match a profile on the DNA database or any other known suspect. Even then it is only used as an investigative tool, to reduce the number of potential suspects when the suspect pool is very large and to help prioritise who to focus on first or next. It is not used as final evidence in court.

For instance, forensic DNA phenotyping might be used to narrow the pool of suspects where investigators already have some idea about where a perpetrator lives (in a cluster of villages, say). One way of doing this is through **familial searching** (described on page 26) to identify potential relatives. If this doesn't generate any leads, then forensic DNA phenotyping might be able to provide clues about the physical appearance of the person they're looking for. The ultimate goal is to identify a group of individuals whose DNA could be sampled (by voluntary screening) and analysed to generate a standard DNA profile, in order to find a match with the DNA profile from the crime scene.

²⁸ Kayser M (2015) Forensic DNA Phenotyping: Predicting human appearance from crime scene material for investigative purposes. *Forensic Science International: Genetics* 18: 33-48

WHO TO PROFILE? FORENSIC DNA PHENOTYPIC TESTS TO NARROW DOWN SUSPECT POOL

If a **DNA profile** is obtained from a crime scene and there is no match on a **national DNA database**, **forensic DNA phenotyping** may be used to predict a suspect's appearance, to *narrow down* from a population to a pool of possible suspects for investigation. Tests can currently predict eg hair colour, sex, eye colour. But these tests aren't 100% accurate, so they cannot (with the exception of sex) be used to *eliminate* suspects. They are still in development, so are rarely used in forensic investigations currently.



Once a suspect pool has been narrowed down to this final group, first DNA samples are taken from each of these individuals. Their DNA profiles are generated and then compared to the DNA profile obtained from the crime scene to see if there is a match. If no match is found the next group up is tested and so on. These groups enable investigators to prioritise which individuals in a population to screen. This is a much more practical and cost effective than simply taking samples from the whole population (which is the alternative approach).

Can DNA tell what the face of a suspected criminal looks like?

It is currently possible to predict eye and hair colour from a DNA sample — although none of these tests are 100% accurate. Some of these tests have been forensically validated (see page 31), and the results of these studies published in scientific journals. Skin colour is likely to be the next appearance trait that forensic scientists will be able to predict from DNA — tests are currently being developed and validated.

See diagram p31

However, knowledge about the genetic basis of any other physical traits is not yet advanced enough for them to be predicted from a DNA sequence. In particular, the genetics of human facial structure is highly complex, and the scientific studies that have been published in this area have identified only a few out of the hundreds or possibly thousands of genes that scientists expect to be involved — each with a very small effect.

This hasn't stopped a USA-based company from marketing a service to reconstruct a face from DNA. The company has not yet published information about its methods (in a peer reviewed journal), nor a validation study which is particularly important in forensic science (see page 29). Yet some police forces have started using these facial reconstruction tests, which has been covered uncritically in the news.

Forensic DNA phenotyping raises some ethical issues too. Whereas standard forensic **DNA profiling** involves **genetic markers** found in parts of the human genome that are not within genes (non-coding regions), the markers used in forensic DNA phenotyping are located within or close to genes involved in the externally visible trait being predicted (coding regions). If forensic DNA phenotyping techniques were extended to also include non-visible characteristics, such as genetic risk of disease, they could reveal personally sensitive, private information, to whoever is doing the testing — which could be of interest to medical insurance companies or certain employers. This can be avoided through regulation, eg in the Netherlands, where externally visible characteristics are legally allowed to be used in forensic DNA phenotyping, but non-visible disease traits aren't.

When DNA is used to make predictions about what a suspect looks like, this will result in estimates of probability and error for each predicted trait, meaning that the weight of this type of evidence can be assessed quite well. It's important that police understand the differences between standard forensic DNA profiling — which can identify individual **DNA profiles** that match -- and forensic DNA phenotyping — which can't identify individuals yet, but provides information that allows individuals to be placed into groups defined by specific visible traits and biogeographic ancestry.



Currently, eye colour, hair colour and skin colour can be predicted reliably and with practically useful accuracy from crime scene DNA, but not yet any other externally visible characteristic.

Manfred Kayser

*Professor of Forensic Molecular Biology, Erasmus MC University Medical Centre, Rotterdam
EUROFORGEN Consortium member*

The face of litter?

Behind every piece of litter is a person who dropped it. So when Hong Kong launched an initiative to tidy up its streets, it tried to shame litterbugs by plastering reconstructions of their faces on billboards across the city. The information for these facial reconstructions came from DNA, which had been retrieved from discarded items such as chewing gum or cigarette butts, and sent to a USA-based company for **forensic DNA phenotyping**.

It made predictions about their sex, hair, skin and eye colour, freckling, biogeographic ancestry, and facial shape. Some of these things can be predicted from DNA, but eg black hair is almost universal within the population of Hong Kong. However, facial shape cannot currently be predicted from DNA because it involves the complex interplay of very many genes (see page 32).

The campaign conveyed the idea that DNA from rubbish could be used to reconstruct what the person who dropped it looks like; that they would be found and prosecuted. The reality is quite different.



Some physical traits, such as skin colour, are closely associated with commonplace understandings of biogeographic ancestry, 'race', or 'ethnic identity'. Because of this, care needs to be exercised in the application of **forensic DNA phenotyping** so that police inquiries are not seen as reinforcing racial stereotyping and perceptions of unequal treatment amongst minority communities.

Robin Williams

*Professor of Forensic Science Studies, Northumbria University, Newcastle-upon-Tyne
EUROFORGEN Consortium member*



The idea that a person's face is reconstructed from DNA traces alone, and the result publicly displayed as a 'photo-fit' to aid police investigations is disconcerting. DNA analysis may be able to predict but cannot determine the actual likeness of a person. However, some may take such images at face value. This could lead to endangering or stigmatising groups of people who may be considered to look similar to such DNA-generated images, even though they are not remotely connected to a crime, or may be innocent.

Matthias Wienroth *Research Fellow in Social Science, King's College London
EUROFORGEN Consortium member*

Predicting biogeographic ancestry

A number of **genetic markers** (known as ancestry informative markers) have been discovered that are much more common among people from some parts of the world (eg Africans) compared to others (eg Europeans). These can be used to predict an individual's biogeographic ancestry, i.e. the broad geographic region their biological ancestors originated from. For now, the available tests that are suitable for crime sample analysis can only reliably predict to which of the major continental groups a person belongs i.e. African, Western Eurasian, East and South Asian, or Native American. They cannot say which country someone comes from.

The terms *ethnicity* and *race* are sometimes used interchangeably with biogeographic ancestry, but strictly, ethnicity reflects a person's social and cultural background, which cannot be detected from DNA. However a person's ethnicity may be strongly associated with their biogeographic ancestry which to some extent can be informed by DNA. The term 'race' refers to a largely outmoded concept of human classification.

DNA tests for refugees by the UK Border Agency

In 2009, the UK Border Agency piloted a scheme to determine the biogeographical ancestry of asylum seekers through DNA testing. It came in response to concerns that people were lying about their country of origin in order to boost their chances of a successful asylum application. Major scientific²⁹ and ethical criticisms led to it being scaled back and then abandoned in 2011. Although DNA tests can predict whether someone's ancestors came from large geographic regions such as continents, such tests cannot predict someone's nationality^{30,31}.

Premature use of forensic DNA phenotyping

Operation Minstead, rapes 1992-2009

When London's Metropolitan Police were struggling to catch a serial burglar and rapist, they turned to a USA-based, DNA testing company to help them establish his ancestral origins via DNA analysis. The company used unspecified ancestry and pigmentation markers to predict that the assailant came from Southern Caribbean regions, so investigators flew to Trinidad. When the perpetrator was finally caught, it turned out he was from Jamaica. Biogeographic ancestry tests can only narrow down to broad geographic regions not specific countries.

²⁹ Balding D, Weale M, Richards M, Thomas M (2010) Genetic and isotopic analysis and the UK Border Agency. *Significance* 7: 58-61

³⁰ Tutton R, Hauskeller C, Sturdy S (2014) Suspect technologies: Forensic testing of asylum seekers at the UK border. *Ethnic and Racial Studies* 37(5): 738-52

³¹ Phillips C (2015) Forensic genetic analysis of bio-geographical ancestry. *Forensic Science International: Genetics* 18: 49-65

However, forensic DNA phenotyping has also been a game changer

Eva Blanco case, 1997

Eva Blanco Puig was a 16-year-old Spanish high school student, who was raped and murdered in Algete, near Madrid in 1997. Police made an application to take DNA samples from men in Algete, including relatives and acquaintances, in the hope of identifying the killer, but the application was turned down. In 2015, semen recovered from Ms Blanco's body was subjected to **forensic DNA phenotyping**, revealing that the perpetrator was likely of North African origin. So investigators narrowed their search, focusing on men from this group who were living in the area in 1997. They took DNA samples from 300 willing volunteers, including two brothers who gave partial matches with conventional **DNA profiling**. This led them to a third brother, Ahmed Chelh, who was arrested and charged with the murder in October 2015³². The case was never heard in court; Mr Chelh was found dead in his cell in January 2016.



The use of new DNA tests to predict ancestry, skin, hair and eye colour reopened this cold case and provided a key shift in focus for the investigators. We were fortunate to have completed reference databases, containing genetic data from many people of North African origin, earlier in 2015 — as well as benefiting from a close and proactive relationship with the investigating team.

Christopher Phillips *Researcher in forensic genetics, University of Santiago de Compostela, Spain EUROFORGEN Consortium member*

All in all, predicting physical appearance from DNA is still in its infancy. But advances in the science are happening so quickly that the investigative and ethical implications need to be considered sooner rather than later.

³² http://elpais.com/elpais/2015/10/04/opinion/1443970167_046935.html (accessed January 2017)

FINAL THOUGHTS FROM EUROFORGEN

DNA profiling is a powerful tool, both for securing criminal convictions, and excluding or exonerating the innocent.

Yet its story is far from complete. Forensic genetics continues to be an innovative, dynamic and evolving field of research, and the amount of information that can be gleaned from the tiniest traces of DNA continues to grow. It is time to take stock of these increased possibilities, and address the challenges that enhanced DNA analysis could bring.

The contributors to this guide have been at the forefront of the development of new types of forensic genetic tests. One strength of the EUROFORGEN Consortium is its multi-national composition and its awareness that different countries have adopted different strategies for processing, interpreting and presenting complex DNA evidence.

As DNA profiling continues to grow more sensitive, and it is used in more investigations, the need for accurate communication between scientists and non-scientists only grows - both to ensure that their expectations of the technology are realistic, and its limits are properly understood. The collaboration between Sense about Science and the EUROFORGEN Consortium aims to improve this process and inform the public about new developments in this exciting field.

USEFUL TERMS TO KNOW

Autosomal DNA

DNA from the 22 pairs of non-sex **chromosomes**, found in the cell nucleus.

Confirmation bias

The tendency to interpret new evidence as confirmation of one's existing beliefs or theories.

Chromosome

The human genome is composed of 23 pairs of chromosomes (46 in total), each of which contain thousands of genes and non-coding DNA.

DNA

DNA is the molecule that carries the genetic information of most organisms including humans.

DNA profile

In the forensic context, this describes the visualisation of the **genetic markers** which have been analysed in an individual's DNA. The most commonly used is an STR DNA profile.

Forensic analysis

Scientific tests or techniques relevant to legal proceedings.

Genetic Marker

Sections of the genome that can have different forms (**alleles**). These sections are highly variable so are chosen to distinguish between individuals. These can be detected in a laboratory and used to generate a **DNA profile**. Every individual has two copies of each genetic marker (because we inherit one version from our mother and one from our father).

Likelihood Ratio

A statistical calculation that summarises the relative support for two hypotheses provided by some evidence. When the likelihood ratio is 1, the evidence available provides equal support for both hypotheses. (Formally, it is the ratio of the probability of the evidence under the two hypotheses).

Locus (plural loci)

A specific, identifiable place in human DNA where there is variability between individuals (**genetic markers** such as **STRs**, **SNPs**).

Mitochondrial DNA (mtDNA)

DNA from mitochondria, which are small energy factories existing in the cell in numerous copies outside of the nucleus. As there are lots of mitochondria in cells their DNA is present in larger amounts and can be more easily detected when DNA from the nucleus is limited or degraded.

Mixed DNA Profile

A DNA profile involving two or more contributors, eg victim(s) and suspect(s).

National DNA Database

Most European countries have national forensic DNA databases storing **DNA profiles** from unsolved criminal cases, as well as from convicted offenders. In a number of countries, such as the UK, DNA profiles from persons arrested but not convicted may also be stored for varying periods of time. In this guide, the term 'DNA database' refers always to these forensic databases.

Nuclear DNA

DNA from the cell's nucleus, which encodes the vast majority of an organism's genes. The most common form of **DNA profiling** involves nuclear DNA (including **autosomal DNA**, Y-chromosomal DNA, but excludes **mtDNA**, that is found outside the cell nucleus).

Partial DNA Profile

An incomplete DNA profile, where some of the **genetic markers** analysed are missing. This can be because the DNA has been degraded by, for example, exposure to heat, water or microorganisms, or because DNA is present at such low levels that accurate marker information cannot be obtained.

Phenotype

The physical characteristics of an individual which are a result of the expression of their genes, as well as environmental factors. **Forensic DNA phenotyping** is the prediction of one or more externally visible aspects of these physical characteristics from the DNA eg eye and hair colour.

Short Tandem Repeats (STRs)

Small sections of DNA (found throughout the human genome), which are made up of short sequences that are repeated. The number of times this sequence is repeated (and hence the length of the section), tends to differ between unrelated individuals and can be measured using STR analysis. This principle forms the basis of the most common types of forensic **DNA profiling** using autosomal STRs (ie those located on the non-sex **chromosomes**). Each STR marker carries two repeats, one inherited from the mother and the other from the father. They are identified by numbers to signify the length of repeat sequences: "11 / 15" or "11 / 11".

Y-STRs are STRs found on the Y chromosome (male only; see below). The other sex chromosome is the X chromosome; females normally carry two X chromosomes, males carry one X and one Y.

Single Nucleotide Polymorphisms (SNPs)

Another form of variability in the DNA of individuals, this time at a single position in the DNA sequence (rather than a repetitive section like with **STRs**).

Y Chromosome DNA

DNA from the Y **chromosome**, one of two sex chromosomes, inherited from father to son so only carried by males. It is found in the cell nucleus.

Royal Statistical Society: Practitioner guides

Four guides intended to assist judges, lawyers, forensic scientists and other expert witnesses in coping with the demands of modern criminal litigation.

<http://bit.ly/2hxZLKa>

Gov.uk DNA guidance

Brings together guidance on DNA published by the Forensic Science Regulator.

<https://www.gov.uk/government/collections/dna-guidance>

Genetics, technology, security and justice: crossing, contesting and comparing boundaries

Six Economic and Social Research Council seminars that critically examine aspects of the contributions of forensic genetics to the production of security and justice in the UK and other contemporary European societies.

<https://www.northumbria.ac.uk/research/academic-departments/applied-sciences/wwwnorthumbriaacukforensicgenetics/>

Matthias Wienroth discusses the series in an ESRC blog. <https://blog.esrc.ac.uk/2016/11/29/geneticstechnology-security-and-justice-the-social-lifeof-dna/#more-1773>

The forensic use of bioinformation: ethical issues

Fingerprinting and DNA profiling are valuable tools in the fight against crime, but there is a debate about whether police powers to keep people's details on record are justified. Nuffield's report (2007) makes recommendations in areas including the use of the National DNA Database.

<http://nuffieldbioethics.org/project/bioinformation/>

Forensic science: A sociological introduction

Christopher Lawless (2016) draws on a wealth of international research and case studies to explore the intersection of science, technology, law and society and examine the production of forensic knowledge.

Inside the cell: The dark side of forensic DNA

Erin Murphy (2016) probes the scientific, statistical, legal, and ethical challenges presented by forensic DNA testing.

Misleading DNA evidence: reasons for miscarriages of justice

Peter Gill (2014) Elsevier. Published under EUROFORGEN funding. The book provides a deep analysis of the Adam Scott, Farah Jama and Meredith Kercher cases, and describes the utility and pitfalls of National DNA databases.

Probability and statistics in forensic science

Isaac Newton Institute for Mathematical Sciences aiming to produce guidelines for reliability estimates for specific forensic techniques

<https://www.newton.ac.uk/event/fos/seminars>

Read our guides at senseaboutscience.org



Find out about EUROFORGEN-NoE research and information resources at www.euroforgen.eu/training/online-resources/

ABOUT US...

The **European Forensic Genetics Network of Excellence** has been operating and developing during five years thanks to the funding of the European Union to bring together forensic scientists, social and legal researchers from nine European countries, who study novel forms of forensic DNA profiling and searching techniques. The **EUROFORGEN** Community will continue to exist in the framework of the International Society for Forensic Genetics.

To achieve a broad public awareness about the advances and key issues in forensic genetics, EUROFORGEN has partnered with **Sense about Science** to produce “Making Sense of Forensic Genetics”. Sense about Science, is an independent campaigning charity that challenges the misrepresentation of science and evidence in public life.

Sense about Science advocates openness and honesty about research findings, and works to ensure the public interest in sound science and evidence is recognised in public discussion and policy making. Sense about Science focuses on socially and scientifically difficult issues where evidence is neglected, politicised or misleading. Sense about Science is a small team working with thousands of supporters, from world-leading researchers to community groups.

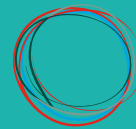
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