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Eureka moment that led to the discovery of DNA fingerprinting

Twenty-five years ago academic Alec Jeffreys stumbled on a remarkable discovery. The scientific breakthrough led to DNA fingerprinting - which has since trapped hundreds of killers, freed the innocent and revolutionised science and criminal justice

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On 10 September 1984, geneticist Alec Jeffreys wrote three words - "33 autorad off" - in his red desk diary. The phrase marked the completion of an experiment, set up that summer, to study how inherited illnesses pass through families. It failed completely.

Yet the project remains one of the most profoundly influential pieces of research ever carried out in a British laboratory, for it produced the world's first DNA fingerprint, a technology that has revolutionised crime scene investigations, led to the convictions of murderers and rapists, and transformed immigration disputes and paternity cases.

Twenty-five years ago the idea that scientists would one day be able to pinpoint an individual from the tiniest trace of their sweat or blood would have seemed laughable. Today we take it for granted. Along with the CCTV camera and the tapping of emails and phone calls, the DNA fingerprint has become part of a civic apparatus that can follow the movements of individuals with unprecedented accuracy.

Thanks to the research by Jeffreys, thousands of dangerous criminals have been caught and imprisoned and thousands of individuals unfairly denied UK citizenship have been allowed to settle in this country. At the same time, millions of individuals have had their profiles stored in databases in Britain, a serious threat to civil liberties according to some organisations and individuals, a point that is - partially - accepted by Jeffreys himself. More than any other modern scientific discovery, DNA fingerprinting raises crucial issues about balancing the use of technology to help society against an individual's right to privacy.

Such concerns were far from the mind of Alec Jeffreys, then a 34-year-old Leicester University genetics researcher, in the summer of 1984. At the time he was seeking ways to trace genes through family lineages and had hit on a fragment of DNA that was repeated on different chromosomes in the cells of men and women.

This genetic stutter could be unique to an individual, Jeffreys realised, and so he devised an experiment to see if he could count those repeats in different individuals and their relatives, as well as in animals such as seals, mice and monkeys.

First, cells were broken open and their DNA extracted. Then this DNA was attached to photographic films. Radioactive probes - which could identify the repeated sections of DNA - were added. Everything was then placed in a photographic developing tank and left over the weekend of 8-9 September. The results, Jeffreys hoped, would reveal ways that might help him study inherited diseases such as cystic fibrosis.

But when he entered his laboratory that Monday morning and removed the film from its tank, he found an odd array of blobs and lines. "My first reaction was 'God, what a mess.' Then I stared a bit longer - and the penny dropped." That piece of film showed a sequence of bars, each representing different numbers of DNA repeats in the various individuals and animals in the experiment.

Crucially, every individual in the sample had a different bar code and could be identified with precision. Jeffreys could even establish kinships: the bands of the DNA supplied by one of his technicians were a composite of her mother's and father's, for example. Even the animal samples showed that individuals could be identified this way. As Jeffreys put it: "It was an absolute Eureka moment. It was a blinding flash. In five golden minutes, my research career went whizzing off in a completely new direction. The last thing that had been on my mind was anything to do with identification or paternity suits. However, I would have been a complete idiot not to spot the applications."

He called his staff together and they began a brain-storming session to find uses for the technology they had stumbled on. Paternity cases were an obvious example, as was the identification of criminals. "But then we thought, how about crime scene samples. Could we get DNA from blood left behind after murders or robberies?"

Today this seems a silly question, attuned as we are to the marvels displayed in CSI Miami and the rest. But in 1984 no one knew how stable DNA was. For all Jeffreys knew, it could break apart rapidly after a cell had died, making crime scene sampling impossible.

"So I spent the next two days cutting myself and leaving blood marks round the laboratory. Then we tested those bloodstains and found that their DNA was intact." Thus the genetics laboratory of Jeffreys was not only the birthplace of DNA fingerprinting; it became the first setting for a DNA crime scene analysis.

Yet the criminal case uses of DNA fingerprinting were not the first to occupy Jeffreys and his team. Its usefulness in immigration cases grabbed immediate attention. A paper about DNA fingerprinting was written by Jeffreys and his team and was published in Nature in March 1985, triggering several newspaper reports. These were instantly followed up by a group of lawyers who were fighting the deportation of a young boy who, said the Home Office, was not the son of a British woman, as she claimed, and had no right to UK nationality.

"In fact, I had never seen the implications for immigration cases," admits Jeffreys. "It was my wife, Sue, who said DNA fingerprinting would make an incredible difference in disputes over nationality. And she was absolutely right."

After talks with the woman's lawyers, Jeffreys agreed to help. However, the case was complicated by the fact that the boy's father was no longer living in Britain and could not be contacted. "It was like a jigsaw with most of the bits missing," says Jeffreys.

Nevertheless he took samples from the mother, her three daughters and the disputed son. The results "blew me away", he recalls. "It was so incredibly simple. When I looked at the film we made of the DNA samples, I could see that every genetic character in the boy was either present in the woman or in his sisters. He was definitely her son."

The Home Office called in Jeffreys and, after a detailed explanation by him, agreed to drop the case. "Afterwards I went over to tell the mother what had happened, that DNA had done its job," says Jeffreys. "She had a bad time for the past two years and it was clearly affecting her health. But the look on her face when I told her, the relief - it was a magical moment. I realised then that we were on to something of real use. We had reached out and touched someone's life."

Over the next decade, DNA fingerprinting was used to test more than 18,000 immigrants who had been refused entry into the UK. Of these, more than 95% produced results that showed they were blood relatives of UK citizens and were therefore entitled to British citizenship - thanks to DNA fingerprinting.

The next two years were "simply insane", adds Jeffreys. He was inundated with calls from families, mostly of Bangladeshi or Pakistani origins, who had been caught up in immigration disputes. "The university switchboard jammed on several occasions with the calls coming into us." A company, Cellmark, was set up in 1987 to take up these cases and took over much of the Jeffreys caseload.

"Then, out of the blue, I got a call from Leicestershire police, who were investigating the rape and murder of two schoolgirls, Linda Mann and Dawn Ashworth, who lived in the village of Narborough outside Leicester." A local man, Richard Buckland, had just confessed to the murder of Dawn but refused to confess to the killing of Linda. Use your DNA fingerprinting technology to prove he killed both girls, they asked him.

So Jeffreys set up his tests, using - in this case - a version of DNA fingerprinting called DNA profiling. Only a limited number of repeated regions are counted, a technique that is quicker to use and requires smaller samples. It was a perfect opportunity to show off the forensic value of genetic fingerprinting, Jeffreys realised, and, as the tests were being completed, he worked through the night to finish them off as quickly as he could. "I just couldn't wait any longer," he says.

He pulled the film from its developing tank. "I had expected to draw a blank and to find there was not enough DNA in the samples of semen that had been taken from the girls' bodies to produce results." He was wrong. The film was covered in black bands, which showed that the semen from both girls came from the same man, but that Buckland's DNA was completely different. He was not the murderer, the tests indicated. "It was a blood-chilling result," adds Jeffreys.

The police's response was terse and Anglo-Saxon. For his part, the geneticist began to worry that the whole concept of DNA profiling was "up the spout" and that there were things going on biologically that science still did not understand. Then the Home Office repeated the tests and produced the same results as Jeffreys. "I was fretting all the time, but they gave me strength," he adds.

In the end, the police accepted Buckland's innocence and on 21 November 1986, at Leicester Crown Court, he was cleared of the girls' murders. Thus the first use of DNA fingerprinting in a criminal case was to help free an innocent man. "I am pretty sure that, given his confession, Buckland would still be in jail today," adds Jeffreys. "Worse, the real perpetrator would have gone on to kill again."

In the end, that perpetrator was caught by a combination of DNA science and "good old-fashioned coppering", as Jeffreys puts it. In January 1987 police asked all local men between 17 and 34 to submit blood for DNA testing in order to eliminate them from their inquiries. By September, 4,000 had provided samples without success - until a chance remark transformed the investigation.

In a pub one day a local man admitted to his mates he had provided blood on behalf of a friend, Colin Pitchfork. One friend told the police, the man and Pitchfork were arrested and the latter's DNA was shown by Jeffreys to match that of the semen from the two girls' bodies. On 23 January 1988 Pitchfork was sentenced to life for the murders of Linda Mann and Dawn Ashworth. "It was the first time on the planet that a criminal investigation had been tackled and solved at a DNA level," says Jeffreys.

Since then, Jeffreys has used DNA profiling to determine a range of intriguing cases. In 1990 he showed that DNA from bones dug up from a Brazilian graveyard by Nazi-hunters was almost certainly that of Josef Mengele, a doctor who had tortured inmates at Auschwitz. A year later, he helped Home Office scientists prove that bones found in a burial pit in Ekaterinburg, 850 miles east of Moscow, were those of the Russian imperial family who had been killed in 1918 during the Russian civil war.

It is a striking body of work, which earned Jeffreys a knighthood in 1994 and which has taken him far from his academic roots and involved him in a startling range of work. He has no regrets, however: "I love it. DNA fingerprinting came out of the blue and turned me round in five minutes flat. There are certain things in science that are historically inevitable, however. I was just lucky that I got to discover DNA fingerprinting. If I hadn't, someone else would have done it by now. I have no illusions about that."

Forensic History

1984 DNA fingerprints are discovered by Alec Jeffreys. At first, these are used extensively to resolve disputed immigration cases.

1987 The first DNA profile is developed, also by Jeffreys. These use pieces of DNA from only a few selected sites on a person's chromosomes. Repetitions of DNA at these sites are counted, producing a set of numbers that acts as a person's DNA identifier. DNA profiles require smaller forensic samples and are quicker to develop. Crucially, they can also be

turned into a sequence of numbers, making it possible for a DNA database to be created.

1995 The UK National Criminal Intelligence DNA Database is established and is used to store the profiles of men and women convicted of crimes in England, Wales and Northern Ireland. A separate database is established in Scotland.

2009 The UK national database is now the largest, per head of population, of any country in the world: almost 10% of the population of England, Wales and Northern Ireland is on the database.

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