Article

# DNA profiling: The first 30 years [Editorial]

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**DNA profiling: the first 30 years**

This year marks 30 years since the seminal publications by Alec Jeffreys et al. appeared in Nature (1-3). At the time Jeffreys was studying gene structure and function; when he found regions of DNA that were highly variable he had the insight to recognize that hypervariable regions could be applied to human identification. On September 10th September 1984 he developed an autorad (which is an X-ray film for those not old enough to remember) with the DNA from his technician and her husband and child that confirmed that the regions of DNA that had been identified were inherited in a Mendelian fashion and so could be used for individual identification and relationship testing.

A striking feature of the development of DNA profiling is the speed that it when from the idea to practice, with the first case, involving an immigration dispute, being resolved with the use of DNA only one year later (4). Its application to criminal casework didn't have to wait much longer when in 1986 it was used to investigate two related murders in Leicestershire, the first one committed in 1983. DNA profiling provided evidence that the two murders had been committed by the same individual and also exclude Richard Buckland who was the main suspect and had confessed to one of the two murders. After a screen of the local population the DNA evidence ultimately identify Colin Pitchfork as the perpetrator. While the Pitchfork case is most closely associated with the early use of forensic genetics Robert Melias bears the distinction of being the first person found guilty in court with DNA being a critical component of the prosecution's case, in November 1987 (5). In today's more regulated climate it's difficult to imagine the transition from the research laboratory to casework and successfully to the court being so rapid.

Whilst the implementation of DNA technology was rapid in the UK, USA and other countries with highly developed forensic science laboratories, it was not without its problems. Technical deficiencies identified in some cases in the USA led to the DNA evidence being successfully challenged, most well known being the Castro case (6, 6). Arguments also rumbled around the methods used to calculate the evidential strength. However, in the face of these problems the use of DNA profiling continued to grow and rapidly became established as on of the

most powerful tools available to forensic science. The evidential strength of DNA evidence has contributed significantly to the refinements around reporting of forensic evidence and also the improvement in quality control/assurance, which has manifested in many countries with the adoption of the international standard ISO 17025.

Of course the technology developed by Alec Jeffreys in 1984 has changed beyond recognition. The methods used for variable number tandem repeat (VNTRs) analysis where challenging; I speak from personal experience here, having used VNTRs when setting up a paternity testing service at the University of Glasgow in the 1990s. Even with improvements in the technology generating a VNTR profile would take days and required a lot of hands-on input from the scientist, and the method was limited in that it needed a relatively large amount of high molecular weight DNA. In the early 1990s more and more was becoming know about the human genome and a large number of short tandem repeats (microsatellites) were identified. These markers had the advantage of being much shorter and therefore more likely to be detected in forensic evidence and could be analyzed using polymerase chain reaction technology (PCR), which developed at around the same time as the first DNA profiles (7, 8). PCR allowed profiles to be generated from much lower amounts of DNA, making possible the profiling of a wide range of biological material recovered from crime scenes. Alec Jeffreys was again at the forefront of applying microsatellite analysis to challenging casework and in 1991 was able to establish the identity of a murder victim in the UK (9) and in 1992 helping to confirm the identity of skeletal remains as being those of Josef Mengele (10).

The first STRs contained dinucleotide repeats and suffered for high levels of stutter and were quickly replaced by tetranucleotide repeats that had lower stutter levels and higher levels of diversity. The development of fluorescent labels allowed multiplex PCRs, to be developed; in the UK this started with a quadraplex and then the second generation multiplex (that incorporated the amelogenin sex test and six STR loci). Further developments to PCR systems adding extra loci, becoming more resistant to PCR inhibitors and being more sensitive have been incorporated and now most laboratories around the world

use PCR systems that analyze 15 STR loci or more along with the amelogenin sex test. The increased power and sensitivity of the tests is welcome, but has also led to extra problems in interpretation as now profiles can be developed from just a few cells, which leads to questions as to how the material came to be deposited at a crime scene as secondary and even tertiary transfer of biological material is a real possibility.

The development of PCR-based systems allowed easy sharing of data between laboratories, which was essential for the development of DNA databases and in 1995 the UK's National DNA Database was initiated. The success of this database in identifying biological evidence recovered from scenes of crime has led to it being copied in many countries. Naturally, the widespread use of DNA profiling has led to further ethical issues focusing around the rights of the state versus those of the individual, and this relationship between the state and individual continues to evolve, with each country developing its own legislation. Worldwide the number of profiles held on DNA databases in 2015 has been estimated to be in the order of 60 million – around half of those in China. This number is only going to increase, with some countries, such as Kuwait, initiating databases that will hold the profiles of all their citizens and residents.

The primary use of forensic genetics remains the analysis of biological evidence recovered from scenes of crimes. However, the expansion of other aspects of DNA profiling are also notable, such as parentage testing, largely assessing paternity. The use of forensic genetics is for the identification of human remains is another area that has continued to develop from the identification of individual bodies to disaster victim identification (DVI) in the mid-1990s (11). Following its success in DVI it has been applied to more complex cases, such as the identification of missing persons in the Balkans following the breakup of the former Yugoslavia. Its use in post-conflict areas continues to grow with large-scale identification projects in several countries and DNA playing a key role.

Thirty years after the first use of DNA profiling in casework the technology and power of the systems are unrecognizable. The 2015 biennial meeting of the International Society of Forensic Genetics has just concluded in Kraków, Poland. The focus has been on emerging technologies such as Next Generation

Sequencing (NGS) that allows massive amounts of data to be generated from trace amounts of DNA, not only allowing identity to be established, but also estimates of age, bio-geographical ancestry and a phenotype (hair, skin and eye colour) to be predicted. The new tools have not been incorporated into routine casework as yet, but will no doubt be used in some contexts. The ability to generate vast amounts of data will present its own problems in terms of evaluating the DNA evidence, presenting it to a court of law, and not least of all ethical issues.

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