

An Introduction to Forensic Genetics

Second Edition

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Foreword

Essentials of forensic science

The world of forensic science is changing at a very fast pace. This is in terms of the provision of forensic science services, the development of technologies and knowledge and the interpretation of analytical and other data as it is applied within forensic practice. Practising forensic scientists are constantly striving to deliver the very best for the judicial process and as such need a reliable and robust knowledge base within their diverse disciplines. It is hoped that this book series will provide a resource by which such knowledge can be underpinned for both students and practitioners of forensic science alike.

The Forensic Science Society is the professional body for forensic practitioners in the United Kingdom. The Society was founded in 1959 and gained professional body status in 2006. The Society is committed to the development of the forensic sciences in all of its many facets, and in particular to the delivery of highly professional and worthwhile publications within these disciplines through ventures such as this book series.

Dr Niamh Nic Daéid

Reader in Forensic Science, University of Strathclyde, Glasgow, Scotland, UK

UK Series Editor

Preface

It is strange to consider that the use of DNA in forensic science has been with us since 1985 and, although a relatively new discipline, it has impacted greatly on the criminal justice system and society as a whole. It is routinely the case that DNA figures in the media, in both real cases and fictional scenarios. The increased interest in forensic science has led to a burgeoning of university courses with modules in forensic science. This book is aimed at undergraduate students studying courses or modules in Forensic Genetics.

We have attempted to take the reader through the process of DNA profiling from the collection of biological evidence to the evaluation and presentation of genetic evidence. Although each chapter can stand alone, the order of chapters is designed to take the reader through the sequential steps in the generation of a DNA profile. The emphasis is on the use of short tandem repeat (STR) loci in human identification as this is currently the preferred technique. Following on from the process of generating a DNA profile, we have attempted to describe in accessible terms how a DNA profile is interpreted and evaluated. In addition, databases of DNA profiles have been developed in many countries and hence there is need to examine their use. While the focus of the book is on STR analysis, chapters on lineage markers and single nucleotide polymorphisms (SNPs) are also provided. A new Chapter has also been added to this edition that provides an overview of DNA profiling of non-human species.

As the field of forensic science and in particular DNA profiling moves onwards at a rapid pace, there are few introductory texts that cover the current state of this science. We are aware that there is a range of texts available that cover specific aspects of DNA profiling and where there is the case, we direct readers to these books, papers or websites.

We hope that the readers of this book will gain an appreciation of both the underlying principles and the application of forensic genetics.

Preface to first edition

It is strange to consider that the use of DNA in forensic science has been with us for just over 20 years and, while a relatively new discipline, it has impacted greatly on the criminal justice system and society as a whole. It is routinely the case that DNA figures in the media, in both real cases and fictional scenarios.

The increased interest in forensic science has led to a burgeoning of university courses with modules in forensic science. This book is aimed at undergraduate students studying courses or modules in Forensic Genetics.

We have attempted to take the reader through the process of DNA profiling from the collection of biological evidence to the evaluation and presentation of genetic evidence. While each chapter can stand alone, the order of chapters is designed to take the reader through the sequential steps in the generation of a DNA profile. The emphasis is on the use of short tandem repeat (STR) loci in human identification as this is currently the preferred technique. Following on from the process of generating a DNA profile, we have attempted to describe in accessible terms how a DNA profile is interpreted and evaluated. Databases of DNA profiles have been developed in many countries and hence there is need to examine their use. While the focus of the book is on STR analysis, chapters on lineage markers and single nucleotide polymorphisms (SNPs) are also provided.

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We hope that the readers of this book will gain an appreciation of both the underlying principles and application of forensic genetics.

1 Introduction to forensic genetics

The development and application of genetics has revolutionized forensic science. In 1984, the analysis of polymorphic regions of DNA produced what was termed ‘a DNA fingerprint’ [1]. The following year, at the request of the United Kingdom Home Office, DNA profiling was successfully applied to casework when it was used to resolve an immigration dispute [2]. In 1986, DNA evidence was used for the first time in a criminal case involving the murder of two young women in Leicestershire, UK: DNA analysis exonerated one individual who had confessed to one of the murders, and following a mass screen of approximately 5000 individuals, identified Colin Pitchfork as the murderer. He was convicted in January 1988 [3].¹

Following on from early success in both civil and criminal cases, the use of genetics was rapidly adopted by the forensic community and now plays an important role worldwide in both the investigation of crime and in relationship testing. The scope and scale of DNA analysis in forensic science is set to continue expanding for the foreseeable future.

Forensic genetics

The work of the forensic geneticist will vary widely depending on the laboratory and country that they work in, and can involve the analysis of material recovered from a scene of crime, kinship testing and the identification of human remains. In some cases, it can even be used for the analysis of DNA from plants [6–18]; animals [19–36] including insects [37–60]; and microorganisms [61–67]. The focus of this book is the analysis of biological material that is recovered from the scene of crime – this is central to the work of most forensic laboratories. Kinship testing will be dealt with separately in Chapter 11 and a brief introduction is given to the testing of non-human material in Chapter 14.

Forensic laboratories receive material that has been recovered from scenes of crime, and reference samples from both suspects and victims. The role of forensic genetics within the investigative process is to compare samples recovered from crime

¹ Note: The first convictions in criminal cases came in 1987, when DNA evidence played an important role in the conviction of two rapists: Robert Melias in the UK and Tommy Lee Andrews in the USA [4, 5].

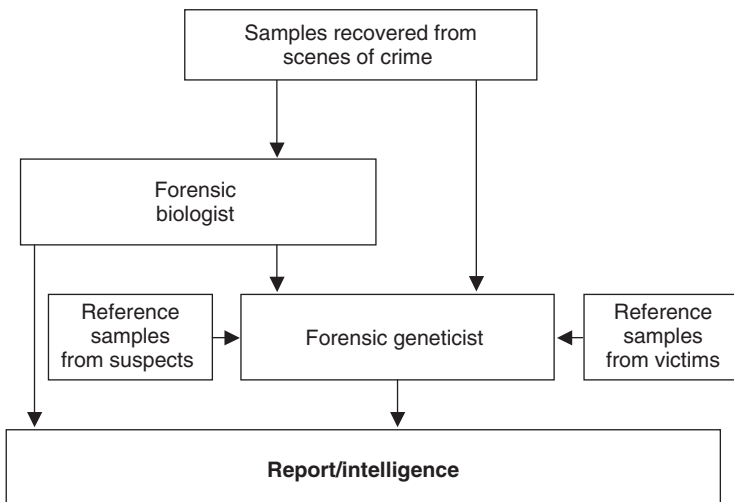


Figure 1.1 The role of the forensic geneticist is to assess whether samples recovered from a crime scene match to a suspect. Reference samples are provided from suspects and also victims of crime

scenes with suspects and possibly victims, resulting in a report that can be presented in court or intelligence that may inform an investigation (Figure 1.1).

Several stages are involved in the analysis of genetic evidence (Figure 1.2) and each of these is covered in detail in the following chapters. In some organizations one person will be responsible for collecting the evidence, the biological and genetic analysis of samples and ultimately presenting the results to a court of law. However, the trend in many larger organizations is for individuals to be responsible for only a very specific task within the process, such as the extraction of DNA from the evidential material or the analysis and interpretation of DNA profiles that have been generated by other scientists, or just reporting the findings.

A brief history of forensic genetics

In 1900 Karl Landsteiner described the ABO blood grouping system and observed that individuals could be placed into different groups based on their blood type. This was the first step in the development of forensic haemogenetics. Following on from this, numerous blood group markers and soluble blood serum protein markers were characterized and could be analysed in combination to produce highly discriminatory profiles. The serological techniques were a powerful tool but were limited in many forensic cases by the amount of biological material that was required to provide highly discriminating results. Proteins are also prone to degradation on exposure to the environment.

In the 1960s and 1970s, developments in molecular biology, including restriction enzymes, Sanger sequencing [68] and Southern blotting [69], enabled scientists to examine DNA sequences. By 1978, DNA polymorphisms could be detected using

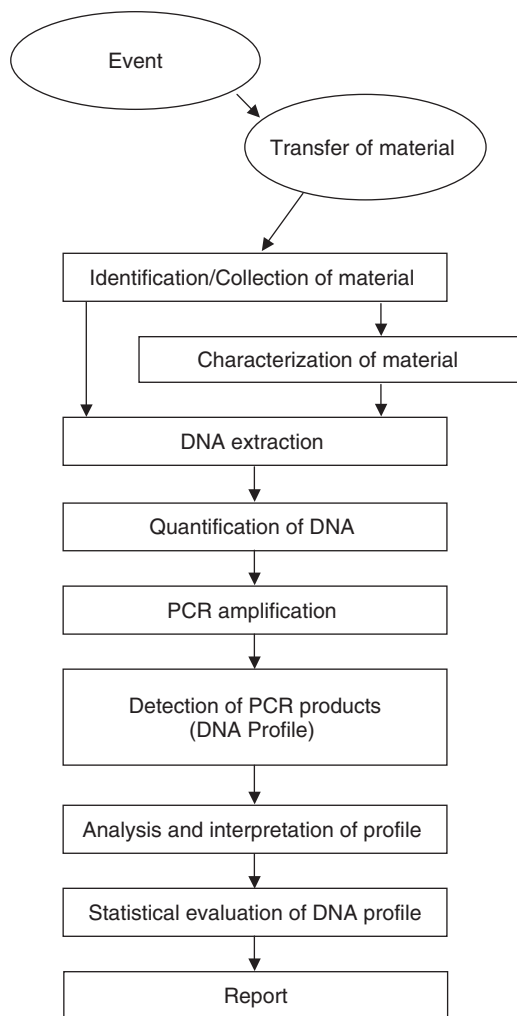


Figure 1.2 Processes involved in generating a DNA profile following a crime. Some types of material, in particular blood and semen, are often characterized before DNA is extracted, allowing the DNA evidence to be linked to a cell type and possible transfer mechanism

Southern blotting [70] and in 1980 the analysis of the first highly polymorphic locus was reported, where the polymorphism was caused by differences in the lengths of the alleles [71].

It was not until September 1984 that Alec Jeffreys realized the potential forensic application of the minisatellite loci [1, 72, 73]. The technique developed by Jeffreys entailed extracting DNA and cutting it with a restriction enzyme before carrying out agarose gel electrophoresis, Southern blotting and probe hybridization to detect the polymorphic loci. The end result was a series of black bands on X-ray film, which was called a DNA fingerprint (Figure 1.3).

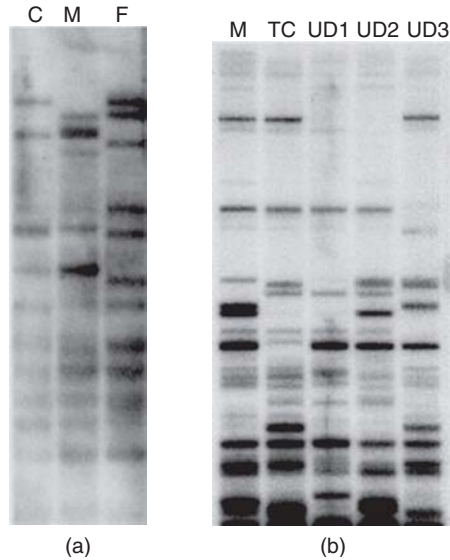


Figure 1.3 (a) The first ever DNA fingerprint, produced in Alec Jeffreys' laboratory on the 10th September 1984. It shows the banding pattern from a mother, child, and father: the bands in the child's profile can be attributed to either the mother or the father. (b) Profiles from a mother, a tested child (TC) (Immigration Officials in the UK did not believe that the boy was the biological son of the mother) and three of the mother's undisputed children (UD 1-3): the results demonstrated that the tested child was indeed the biological son of the woman and was therefore allowed to stay in the UK [2]. (Images provided by Sir Prof Alec Jeffreys, Department of Genetics, University of Leicester, UK)

With the first DNA fingerprints the multi-locus probes (MLPs) detected several minisatellite loci simultaneously, leading to the multiple band patterns. While the multi-banded fingerprints were highly informative they were difficult to interpret. New probes were designed that were specific to one locus (single locus probes, SLPs) and therefore produced only one or two bands for each individual [74] (Figure 1.4).

Minisatellite analysis was a powerful tool but suffered from several limitations: a relatively large amount of DNA was required; it would not work with degraded DNA; comparison between laboratories was difficult; and the analysis was time consuming. Even so, the use of minisatellite analysis, using SLPs, was common for several years [75] until it was replaced by polymerase chain reaction (PCR)-based systems.

A critical development in the history of forensic genetics came with the advent of a process that can amplify specific regions of DNA – the PCR (see Chapter 5). The PCR process was conceptualized in 1983 by Kary Mullis, a chemist working for the Cetus Corporation in the USA [76]. The development of PCR has had a profound effect on all aspects of molecular biology including forensic genetics, and in recognition of the significance of the development of the technique Kary Mullis was awarded the Nobel Prize for Chemistry in 1993.

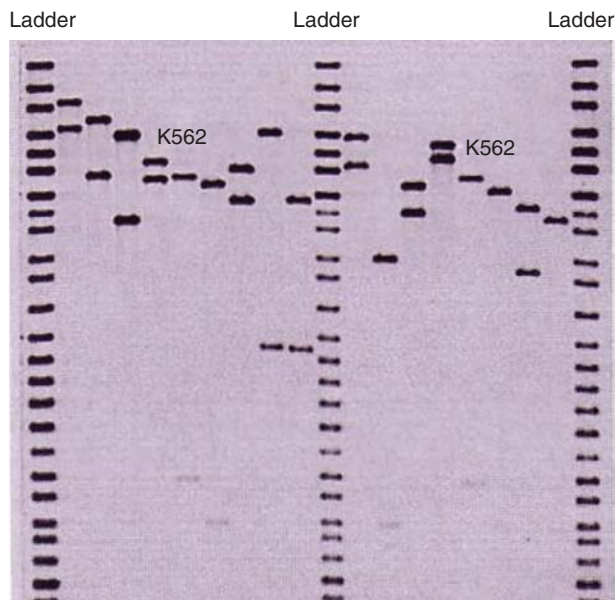


Figure 1.4 Minisatellite analysis using a single locus probe: ladders were run alongside the tested samples that allowed the size of the DNA fragments to be estimated. A control sample labelled K562 was analysed along with the tested samples

The PCR increases the sensitivity of DNA analysis to the point where DNA profiles can be generated from just a few cells, reduces the time required to produce a profile, can be used with degraded DNA and allows just about any polymorphism in the genome to be analysed.

The first application of PCR in a forensic case involved the analysis of single nucleotide polymorphisms in the human leukocyte antigen (HLA)-DQ α locus (part of the major histocompatibility complex (MHC)) [77] (see Chapter 12). This was soon followed by the analysis of short tandem repeats (STRs), which are currently the most commonly used genetic markers in forensic science (see Chapters 6–8). The rapid development of technology for analysing DNA includes advances in DNA extraction and quantification methodology, the development of commercial PCR-based typing kits and equipment for detecting DNA polymorphisms.

In addition to technical advances, another important part of the development of DNA profiling that has had an impact on the whole field of forensic science is quality assurance. The admissibility of DNA evidence was seriously challenged in the USA in 1989 – *People v. Castro* [78]; this and subsequent cases in many countries have resulted in increased levels of standardization and quality assurance in forensic genetics and other areas of forensic science. As a result, the accreditation of both laboratories and individuals is an increasingly important issue in forensic science. The combination of technical advances, high levels of standardization and quality

assurance have led to forensic DNA analysis being recognized as a robust and reliable forensic tool.

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