

# 1 Introduction to forensic genetics

Over the last 20 years 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 a real case, when it was used to resolve an immigration dispute [2]. Following on from this, in 1986, DNA evidence was used for the first time in a criminal case and identified Colin Pitchfork as the killer of two school girls in Leicestershire, UK. He was convicted in January 1988. The use of genetics was rapidly adopted by the forensic community and plays an important role worldwide in the investigation of crime. Both the scope and scale of DNA analysis in forensic science is set to continue expanding for the foreseeable future.

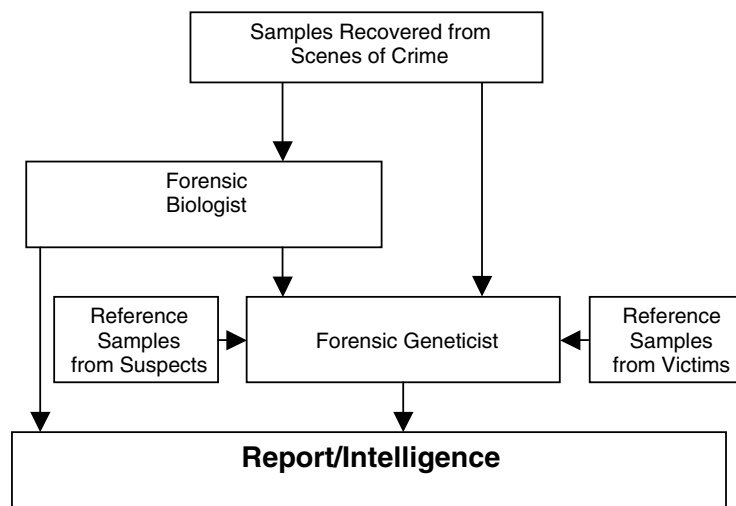
## Forensic genetics

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, paternity testing and the identification of human remains. In some cases, it can even be used for the analysis of DNA from plants [3, 4], animals [5, 6] and microorganisms [7]. 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.

Forensic laboratories will 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 scenes with suspects, resulting in a report that can be presented in court or intelligence that may inform an enquiry (Figure 1.1).

Several stages are involved with 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



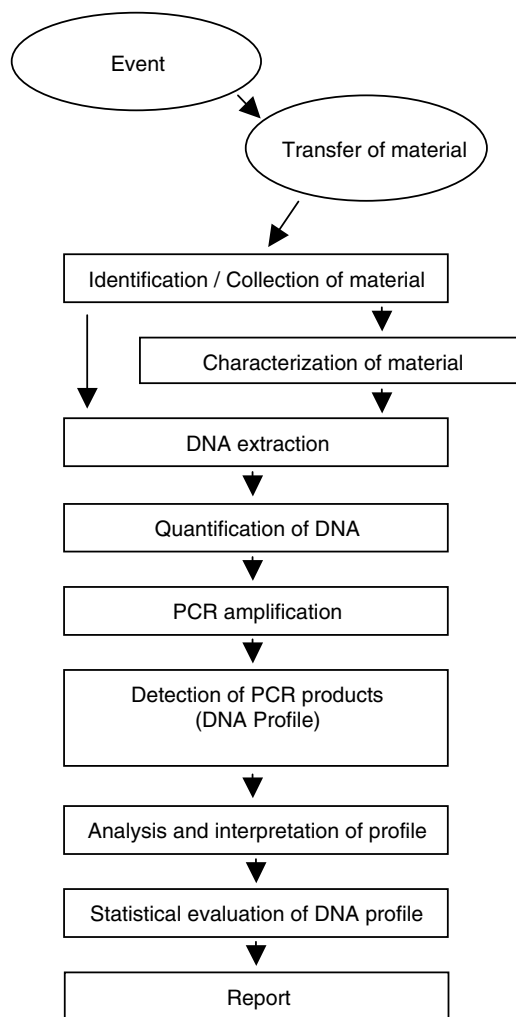
**Figure 1.1** The role of the forensic geneticist is to assess whether samples recovered from a crime scene match to a suspect. Reference samples are provided from suspects and also victims of crime

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.

### 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. In 1915 Leone Lattes published a book describing the use of ABO typing to resolve a paternity case and by 1931 the absorption–inhibition ABO typing technique that became standard in forensic laboratories had been developed. 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.

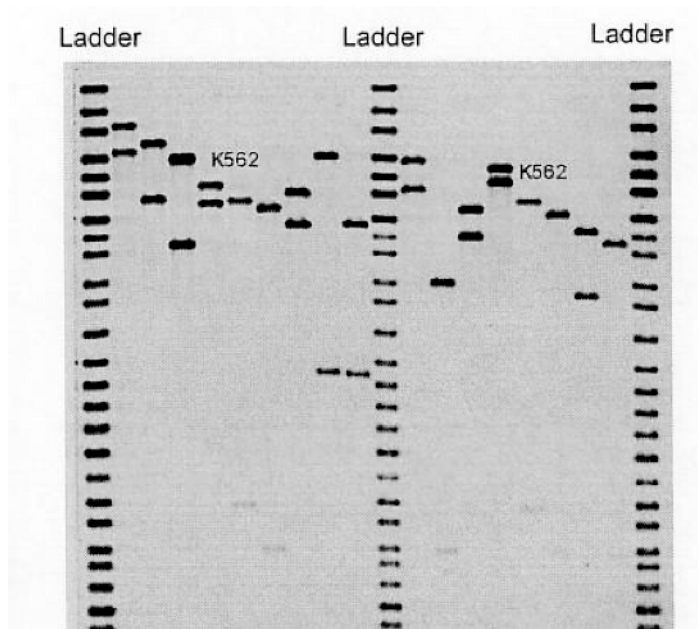
In the 1960s and 1970s, developments in molecular biology, including restriction enzymes, Sanger sequencing [8], and Southern blotting [9], enabled scientists to examine DNA sequences. By 1978, DNA polymorphisms could be detected using Southern blotting [10] and in 1980 the analysis of the first highly polymorphic locus was reported [11]. It was not until September 1984 that Alec Jeffreys realized the potential forensic application of the variable number tandem repeat (VNTR) loci he had been studying [1, 12]. The technique developed by Jeffreys entailed



**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

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 (Figure 1.3). VNTR 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.

A critical development in the history of forensic genetics came with the advent of a process that can amplify specific regions of DNA – the polymerase chain reaction (PCR) (see Chapter 5). The PCR process was conceptualised in 1983 by Kary Mullis, a chemist



**Figure 1.3** VNTR 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 is analysed along with the tested samples

working for the Cetus Corporation in the USA [13]. 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 PCR, Kary Mullis was awarded the Nobel Prize for Chemistry in 1993. The PCR increased the sensitivity of DNA analysis to the point where DNA profiles could be generated from just a few cells, reduced the time required to produce a profile, could be used with degraded DNA and allowed 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  $DQ\alpha$  locus [14] (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 to 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 control. The admissibility of DNA evidence was seriously challenged in the USA in 1987 – ‘*People v. Castro*’ [15]; this case and subsequent cases have resulted in increased levels of standardization and quality control 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 control have led to forensic DNA analysis being recognized as a robust and reliable forensic tool worldwide.

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