

# DNA Forensic and Forensic Investigative Leads

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## Abstract

The chapter explores the fundamental principles of Deoxyribonucleic acid (DNA) in the context of forensic DNA profiling. Moreover, it explores the use of forensic DNA databases and the comparative searches carried out within them. The outcomes of these comparative searches produce Forensic Investigative Leads (FILs), which require communication with detectives for more investigation and follow-up. To successfully investigate these FILs, it is crucial to have efficient communication and engage in multidisciplinary detective work. This entails examining further corroborating evidence that could either absolve an innocent person from blame or contribute to the apprehension and compelling conviction of the perpetrator.

**Keywords:** Deoxyribonucleic acid • Forensic DNA database • Familial searching • Forensic investigative leads • Forensic investigative genetic geology • Criminal casework investigations

## Introduction

Criminal investigations extensively depend on Deoxyribonucleic Acid (DNA) forensic investigation leads, transforming forensic science by offering crucial information for identifying suspects, connecting individuals to crime scenes and verifying or disproving criminal testimonies [1,2]. Although the act of discarding a cigarette butt in a public area may appear harmless, justifying the presence of DNA discovered in a recently burglarised residence presents a substantial difficulty for individuals who are being investigated [3].

### Deoxyribonucleic Acid (DNA)

DNA, the hereditary code of all living beings, is crucial in distinguishing individuals based on biological material by examining genetic variations [4-6]. DNA, which encodes biological properties such as protein expression and physical attributes, shows significant similarities among individuals, with just a 0.5% variation in genetics [7]. Genetic information plays a fundamental role in forensic science in confirming identity and conducting criminal investigations [4,8,9].

Different patterns of repeating sequences found in DNA, such as VNTR and STR, allow for the unique identification of individuals using tandem repeat analysis [10,11]. Despite having the same genetic material, monozygotic twins have separate genetic profiles due to mutations that occur after the division of the zygote, which sets them apart [12]. Despite their widespread use, conventional length-based STR approaches cannot distinguish between monozygotic twins [4,13-15]. Ultra-deep Multiple Parallel Sequencing (MPS) has emerged as a solution for finding genomic variations, such as copy number differences and single nucleotide polymorphisms [16-18].

In creatures such as humans, DNA is found in nuclear DNA in cell nuclei

and mitochondrial DNA(mtDNA) in mitochondria. In the double helix structure of DNA, the nucleotides are coupled together - adenine with thymine and guanine with cytosine - to create base pairs [19,20]. The process of DNA replication and recombination, essential for genetic inheritance, is made possible by the complementary base pairing. During fertilisation, chromosomal pairs are separated, where each parent contributes half of the DNA to the offspring [21,22]. This separation leads to the creation of genetic variety. DNA replication, which is susceptible to mutations, results in the emergence of genetic variants as time progresses, which occur due to errors in DNA replication or repair, resulting in alterations in DNA sequences [23,24]. These changes contribute to the distinctiveness of individuals [25,26]. Forensic DNA profiles employ fragments from locations in the genome to detect alleles and heterozygosity by analysing repetitive nucleotide sequences [2,27,28]. Different alleles at each locus are distinguished by motif repetition and sequence type variations [29].

### Forensic DNA profiling

Forensic DNA profiling was first introduced in the early 1980s [30]. Since then, it has been developed and established as a powerful tool. The advancements in DNA analysis have significantly improved the criminal justice system by enabling the investigation of small biological traces such as blood, saliva and skin cells to link perpetrators [31,32]. Forensic Identification Leads (FILs) play a crucial role in connecting persons to crime scenes and verifying or disproving reports of incidents [2]. In addition, a perpetrator's DNA on objects such as balaclavas can indicate an effort to hide one's identity while engaging in illegal actions [32].

Forensic DNA profiling became possible due to identifying notable discrepancies in minisatellite repeats among people [4, 33]. Southern Blotting techniques were initially used to generate distinct DNA patterns by exploiting minisatellite dispersion. This technique used restriction enzymes to generate DNA pieces of different sizes [33]. The approach, introduced by Sir Alec Jefferys in 1984, resulted in the initial successful DNA profiling in the U.K. This technology has been essential in criminal investigations and in proving innocence [34,35].

The Polymerase Chain Reaction (PCR) significantly impacted DNA analysis by enabling the amplification of small quantities of DNA [36]. The Polymerase Chain Reaction (PCR) is a DNA analysis technique that amplifies Short Tandem Repeat (STR) loci. This process produces amplicons labelled with fluorescent markers, allowing for the determination of their size. The amplicons are separated based on size using either gel or capillary electrophoresis, resulting in the generation of a STR genotype that can be compared. The PCR technique and improvements such as qualitative PCR

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