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Forensic DNA Profiling: Its Role and Advancements in Criminal Investigations

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Abstract—*DNA profiling, as it is known today, basically revolutionized forensic science to a degree of being capable of establishing whose identity was matched with or linked to the crime scene. Since its use the first time in the 1980s, it has become an indispensable tool in criminal investigations that led to the conviction of many criminals and to the exonerating many wrongly accused persons. This review examines the fundamental techniques involved in forensic DNA profiling, which include STR analysis, PCR, mtDNA testing, and NGS. This article further covers the emerging role of DNA databases and investigative genetic genealogy in the solve of cold cases and mass disaster identifications. Moving forward, we look into new breakthroughs in DNA phenotyping, gene editing such as CRISPR technology, and AI in DNA analysis. As DNA forensic technology advances, its use remains at the forefront for bettering justice and criminal investigations*.

Keywords— *DNA profiling, Forensic Genealogy, Genetics, Crime scene investigation, PCR analysis, STR analysis*.

I. INTRODUCTION

The Emergence of DNA Profiling in Criminal Investigations

DNA Profiling is one of the most important developments in the area of forensic science. It enables reliable identification and linking of suspects to a crime scene. Since the first application of DNA profiling in the 1980s, DNA profiling dramatically changed the course of many investigations concerning crimes while providing a way of collecting evidence backed by hard scientific proof (1) (2). It was by the evolution of techniques such as STR analysis and PCR amplification that forensic scientists were able to work even with samples of minute amounts of biological material, with unprecedented accuracy in identifying who a sample belonged to. DNA profiling has come in use not only to help convict the guilty but also to exculpate the wrongly accused, notably through work from organizations like The Innocence Project (3, 4).

Introduction to DNA

Deoxyribonucleic acid (DNA) is the hereditary material in humans and most other organisms. It carries genetic instructions essential for the development, functioning, and reproduction of all living beings. DNA's double-helix structure, consisting of four nucleotide bases—adenine, cytosine, guanine, and thymine—forms the blueprint for an individual’s genetic identity (5). DNA technology allows forensic scientists to compare biological samples from crime scenes to known DNA profiles with remarkable accuracy (3). By analyzing unique patterns in an individual's DNA, forensic biologists can distinguish between individuals, except for identical twins who share the same genetic code.

Historical Development of DNA Technology in Forensics

The first major application of DNA to forensic science was led in early 1980s by Sir Alec Jeffreys, who developed DNA fingerprinting (1). It was a discovery that led to the first case of DNA-based convictions for the murders of two girls in 1986 from the United Kingdom. From there on, forensic DNA technology has dramatically improved, with some other great steps forward including STR analysis, PCR, and mitochondrial DNA sequencing (6). For years, DNA became very important for identification of suspects and victims when committing a crime. DNA databases continue to expand, making DNA more useful in solving cold cases and mass disasters and much more. (7).

Fundamentals of DNA Technology in Forensics

Structure and Function of DNA

DNA is a molecule that consists of two chains coiled into a double helix, containing all the instructions required to create and operate organisms. There exists a unique DNA sequence in every human being; therefore, the difference between two individuals is only possible through the change in specific regions while it is applied to forensic science. These unique strands are mainly gathered by forensic investigators to create a DNA profile that will be matched with samples obtained from various crime scenes. Since DNA is known to be stable for an incredibly long time, it can be retrieved even when evidence is old or degraded, which makes it play an extremely important role for law enforcers (5).

Types of DNA Used in Forensic Science

Forensic scientists take advantage of two types of DNA; namely the nuclear DNA and the mitochondrial DNA. Nuclear DNA is located within the nucleus of the cell and contains the complete set of genetic guidelines passed down by both parents. Mitochondrial DNA, on the other hand, is found in the mitochondria and is passed down through only the mother. While nuclear DNA provides the full genetic

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profile, mtDNA is utilized when nuclear DNA degraded or it is not found. In fact, it is useful at the point where sample collected from the scene of crime is either poor in quantity or degraded in quality. The usage of nuclear or mitochondrial DNA depends on the quality and quantity of sample collected from the crime scene (8, 9).

Sources of DNA Evidence

DNA can be isolated from many biological samples found in contaminated areas. Some of the most common are blood, saliva, hair, skin cells, semen and sweat. Even very minute amounts of DNA have become usable through forensic analysis since they are amplified by the process of PCR technology. DNA can also withstand numerous forms of environmental conditions, thereby making it feasible to obtain genetic evidence long after a crime has been committed (10).

DNA Analysis Techniques in Forensics

Short Tandem Repeat (STR) Analysis

Of all the techniques of forensic DNA profiling, the most widely used is Short Tandem Repeat (STR) analysis. STR is simply a series of repetitive DNA sequences that vary greatly between individuals, making them suitable for identification. A forensics scientist has the ability to make a positive identification match between STR markers from a crime scene sample and a known DNA profile with near certainty. The traditional panel of STR loci used in forensic analysis is identical across all international databases, thereby improving the ability to compare profiles across jurisdictions (16).

Polymerase Chain Reaction (PCR)

One such method used for amplifying small or degraded DNA samples is PCR, which makes trace amounts of DNA at the crime scene analyzable. Through PCR, forensic scientists can develop a profile on any amount of evidence and copy billions of portions of DNA, heightening their expectations to seek assistance in solving crimes. This technique has been of great help in those samples where DNA is scanty, degraded, or otherwise contaminated. As a result, forensic analysts will still be able to obtain usable information therefrom (17, 18).

Mitochondrial DNA (mtDNA) Testing

Mitochondrial DNA (mtDNA) is used in a case where nuclear DNA does not exist or is degraded and cannot be used in producing a good and reliable result. Mitochondrial DNA (mtDNA) is more resistant to degradation from the environment due to its circular nature and high copy number in cells. Forensic scientists rely mostly on mtDNA when dealing with issues of skeletal remains, hair shafts, or highly degraded samples. However, since mtDNA is transmitted maternally, it lacks the degree of individual specificity characterizing nuclear DNA, therefore, finds it better suited for identifying familial relationships (19, 20).

Y-Chromosome Analysis

Y Chromosome testing traces back male lineage using the specific Y-STR markers on the Y chromosome, that are passed straight down from father to son through all generations. The method is particularly useful in distinguishing between male and female DNA in sexual assault cases or cases of kinship relations. Y-STR testing is one of the best practicable techniques in such cases because it can help forensic scientists to provide pure male DNA samples for further analysis (21).

Next-Generation Sequencing (NGS)

Next-generation sequencing practice is nowadays becoming widely promoted in forensic science. With NGS, whole-genome or whole-region DNA sequencing becomes possible-meaning a far more comprehensive profile of one's genetic makeup than possible with traditional methods. As a consequence, current forensic scientists can actually have better resolution in DNA analysis, especially with regards to complex mixtures and highly degraded samples. It may further provide an investigator with ancestry, phenotype, and other genetic characteristics that may be useful in solving a case (22, 23).

DNA in Criminal Investigations: Applications

Identification of Suspects

One of the extensive applications of DNA in criminal investigations is suspect identification by matching DNA profiles to crime scene evidence. DNA profiling has been very helpful in solving cases of burglary, violent crimes, and many other cases by linking biological materials directly to suspects. Given the credibility of DNA testing, it becomes a very important tool for investigations during which various other forms of evidence, including testimonial evidence, may not be credible. DNA fingerprinting has been known to be a technique that can successfully exculpate or incriminate culprit. Additionally, the increasing forensic DNA databases, helps the law enforcement agencies to search instantly for matches in the DNA and bring repeat offenders to justice (3).

Exoneration of Wrongfully Convicted Individuals

DNA testing is very effective in disproving many false positives. Organizations like The Innocence Project have contributed immensely towards this. Since its inception, hundreds of people who were wrongly imprisoned for crimes they did not commit have been exonerated using DNA evidence. In most cases, DNA evidence based on the application of modern DNA technology led to a real attacker being identified as the perpetrator in the crime committed against a victim; therefore, such cases prove that justice indeed triumphs with the existence of DNA evidence. DNA evidence applied exoneratively increased sensitivity toward the possibility of miscarriages of justice in cases that relied heavily upon nonevidence-based facts (24) (25).

Cold Case Investigations

Many cold cases were opened and solved due to advances in DNA technology over the years. As national DNA databases grew and techniques to recover DNA improved, this allowed investigators to go back into older evidence and tie it to suspects. Cold cases that were solved to the very surprise of people, as they had been arrested due to the Golden State Killer match, were found with aged crime scene DNA matched to profiles found within public genealogy databases (26). DNA technology still gives cold case investigations new life and a sense of hope for justice in

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cases previously considered unsolvable. Currently, individuals who are previously known to the investigating authorities can be identified by forensic DNA analysis (27) (28).

Mass Disasters and Missing Persons Identification

In the case of mass disasters, like plane crashes or natural disasters, DNA is incredibly relevant for identifying casualties. After a DNA profile of the victims is compared with that of relatives, forensic scientists may prove extremely useful in bringing closure to grieving families. Where other methods, like dental or fingerprint records, are not available or not useful, DNA technology cannot be substituted. Missing persons cases, on the other hand, and DNA databases really are a source of great value in matching unidentified dead people with missing persons cases (29) (30, 31).

DNA Databases and Their Role in Investigations

National DNA Databases

National DNA databases are currently among the most effective resources for criminal investigators looking for information about unidentified offenders. Currently, European databases hold about six million DNA profiles from both convicted and unconvicted criminals. Additionally, over a million person-to-stain and stain-to-stain hits have been obtained. The databases' diverse organizational structures are based on each country's unique regulatory framework (32).

Investigative Genetic Genealogy

It is a new approach called investigative genetic genealogy, where the genetic data is sourced freely on genealogy websites to crack cases. In the past couple of years, this technique has been very instrumental in solving multiple high-profile cold cases, including the one on the Golden State Killer (26). The DNA Doe Project has identified nine other cold cases (33). However, this effective tool raises ethical issues concerning privacy and personal genetic information used in criminal investigation. The balance of the use of genetic data that crime solving stands against public trust remains one of the highly contentious debates ever (34).

Ethical and Privacy Concerns

These advances have been linked to several ethical implications, including privacy issues, consent, and the potential misuse of genetic data. Among the reasons advanced for the ethics debate are claims that the DNA profiles are infringing civil liberties in the collection and storage of DNA, and such concerns ought to be taken into consideration against the need for a balance between offenders who have not been convicted to being punished and individuals who may victimize others. All these considerations raise the importance of severe regulations and proper oversight of the application of forensic DNA technologies (35) (36).

Challenges in Forensic DNA Analysis

Contamination and Degradation of DNA Evidence

DNA samples are fragile and can be contaminated and degraded. Still, that is such a small percentage in comparison to the fact of degradation of integrity concerning the forensic investigations. The DNA samples have environmental influences such as heat, humidity, and time that gradually degrade them; in the analysis, it becomes very difficult. Besides that, false positives or inconclusive results can arise from contamination or cross-contamination when collecting at the crime scene (37). This calls for the setting up of forensic laboratories under strict controls to prevent contamination, thus ensuring validity in results on DNA (38) (39).

Interpretation of Mixed DNA Samples

One of the common challenges forensic scientists faces when dealing with DNA samples is the potential presence of DNA from more than one individual. Essentially, the concept of mixing DNA profiles involves techniques and software more advanced than deconvolution in making any interpretations of data accrued and associating DNA with people (40). In some instances, the mix sample may be just too complex to make any concrete findings, and sometimes turns out to be problematic in court (41). Actually, the application of mixed DNA samples requires critical accuracy in their interpretation to avoid misuse of DNA evidence during criminal cases (42).

Legal and Scientific Challenges

Application of DNA evidence in the courtroom demands that strict legal and scientific standards must be followed. The DNA evidence analysis should meet either Daubert or Frye standards in any jurisdiction to be admitted to the court. The forensic experts should be able to apprise the judges and the juries on how the methodology works, what reliability it has, and what is possible in limitations while using this DNA evidence (43).

Limitations of DNA Technology

DNA technology may not be infallible. Sometimes DNA evidence may become too degraded, contaminated, or insufficient for analysis. Furthermore, DNA alone can neither prove a person guilty nor innocent as DNA identifies a person only to a particular place or object and not even about his involvement in the crime (3).

The Future of DNA in Forensics

Advances in DNA Phenotyping

DNA phenotyping is a relatively new technology where specific genetic information is used to predict an assortment of physical traits, including hair and eye color, as well as ancestry (44). Such technology might identify unknown suspects when other methods of traditional DNA profiling could not identify a suspect. However, using phenotyping technology in criminal investigations raises race-profiling issues (45, 46).

Use of CRISPR and Gene-Editing in Forensic Science

It is known for gene editing, and it could revolutionize forensic science. With applications at still very early stages, it conceivably can be used in the future to enhance recovery of DNA or even attempt to reconstruct damaged genetic material. The use of gene-editing technology in forensic science involves a host of ethical issues which thus will necessitate careful consideration (47, 48).

Epigenetics in Forensic Science

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Growing interest in forensic science also occurs in the area of epigenetics, which are studies that help in the development of change in the gene expression. DNA methylation patterns are used to understand aspects about the age and lifestyle and overall exposure of an individual (49). This in turn may be more detailed in the construction of suspect or victim profiles (50).

Automation and AI in DNA Analysis

The future of forensic DNA analysis is in the automation of laboratory processes and using AI that interprets complex data. Processing times can be significantly reduced because the analysis of DNA mixtures can more precisely be given and yields a better result. It is envisioned that the entry of AI and automation into forensic DNA laboratories will catalyze faster and more reliable results (51).

II. Conclusion

DNA technology has triggered the forensic science sector, one that enables police to deal with their evidences a lot more powerfully in detecting suspects and solving cold cases and exonerating the innocents. From STR analysis to Next-Generation Sequencing, new opportunities have emerged in forensic profiling, turning cases around. DNA profiling has been one of the most vital components of modern criminal investigations, ensuring that justice is carried out with the highest level of scientific accuracy. Through DNA evidence in courtrooms, criminal prosecutions have been improved in terms of reliability and public trust in the legal system has therefore been increased. Forensic DNA technology will increasingly play a role in criminal investigations. Some of the emerging techniques that are on the horizon include DNA phenotyping, CRISPR, and AI-driven analysis, all of which promise a great future ahead. This would give forensic scientists greater details and accuracy to forward to law enforcement agencies.

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