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From Genes to Justice: Ethical Dilemmas and Scientific Advancements in DNA Profiling for Legal Cases

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ABSTRACT

The utility of DNA in humans is continually expanding as scientific research advances, leading to new discoveries and applications across various fields. DNA, or deoxyribonucleic acid, serves as the genetic blueprint for all living organisms, including humans. For long it has being used for genetic information, treatment of genetic disorders, pharmaceutical research, biotechnology research, paternity test and cancer research/ treatment. DNA profiling has emerged as a powerful tool in forensic science, revolutionizing the field of criminal investigations and legal proceedings. The DNA profiling and its pivotal role in establishing individual identity, elucidating biological relationships, and aiding law enforcement agencies in solving crimes. India enacted the landmark bill called The DNA Technology (Use and Application) Regulation Bill, 2019 and with this it opened many areas of research which Indian forensic scientist could not do due to ethical issues and controversies surrounding its acceptability in legal proceedings. This paper will try to study the act and vast possibilities of DNA Profiling and Genealogy in legal proceedings.

Keywords: DNA, DNA profiling, DNA Genealogy, DNA Databank.

I. INTRODUCTION

The United Kingdom has been a pioneer in the use of DNA profiling in legal proceedings. DNA evidence is widely accepted in both criminal and civil cases.[1] The National DNA Database (NDNAD) in the UK is one of the largest DNA databases globally and is extensively used in criminal investigations. Along with them almost all developed countries including USA, European countries, China, Canada, Australia and many more have been extensively using DNA profiling for the legal proceedings.

Till date India faced ethical and political dilemma in opening this area for legal proceedings. The DNA Technology (Use and Application) Regulation Bill, 2019, represents a landmark

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legislative initiative in India, poised to significantly augment the landscape of DNA profiling and research in the country. This bill will help to regulate the use of DNA technology, providing a structured framework for its application in various areas, including forensic investigations, identification of missing persons, and research endeavours.[2]

The bill's will lead to the establishment of the National DNA Data Bank and Regional DNA Data Banks and hence will help establishment of extensive genetic databases. These will serve investigative purposes and also open up avenues for large-scale genetic studies. Researchers can harness the wealth of genetic information within these databases to conduct population studies, explore genetic markers for diseases, and contribute to the global understanding of human genetics.[2]

Let's study the DNA profiling and various techniques to achieve maximum success in legal proceedings.

II. DNA PROFILING

Deoxyribonucleic acid (DNA) profiling, a revolutionary advancement in molecular biology, has transformed the landscape of forensic science, genetics, and medical diagnostics. With its unparalleled precision and discriminatory power, DNA profiling has transcended its initial forensic applications to become a cornerstone in various domains, shaping legal proceedings, unravelling genetic mysteries, and influencing medical decision-making.[3]

DNA profiling, commonly known as DNA fingerprinting, relies on the unique genetic blueprint inherent in every individual's DNA. The technique involves the extraction, amplification, and analysis of specific regions of the DNA, unveiling distinct patterns that serve as a genetic signature.[4] Originally conceived as a tool for criminal investigations, DNA profiling has evolved to encompass a spectrum of applications, from resolving paternity disputes and identifying missing persons to advancing our understanding of human genetic diversity.

The historical trajectory of DNA profiling is marked by seminal milestones, including the advent of polymerase chain reaction (PCR) techniques, the discovery and utilization of short tandem repeats (STRs), and the contemporary integration of high-throughput sequencing technologies.[4] These technological strides have not only enhanced the efficiency and accuracy of DNA profiling but have also broadened its scope, enabling applications in fields as diverse as personalized medicine, population genetics, and evolutionary studies.

In the forensic arena, DNA profiling stands as an invaluable tool for criminal investigators, aiding in the identification of perpetrators, exclusion of innocent suspects, and establishment of

links between crime scenes and individuals. The establishment of national DNA databases has further amplified its impact, facilitating cross-border collaborations and contributing to the global effort to combat crime.

Beyond forensics, DNA profiling plays a pivotal role in family law, allowing for the determination of paternity with unprecedented accuracy. Its value in medical field is huge as it will help study the patient in tailor made fashion whereby every individual genetic makeup could be studied and hence we can optimize therapeutic outcomes and minimize adverse effects.

III. HISTORICAL BACKGROUND ON DNA PROFILING TECHNIQUES

The foundation of DNA profiling lies in the elucidation of the DNA molecule's structure by James Watson and Francis Crick in 1953.[5] Their discovery, based on the work of Rosalind Franklin and Maurice Wilkins, revealed the iconic double-helix structure of DNA, unlocking the molecular code governing heredity.[5]

The 1970s witnessed the advent of restriction enzymes, proteins capable of cutting DNA at specific sequences. This discovery, coupled with the development of recombinant DNA technology, allowed scientists to manipulate and analyze DNA with unprecedented precision. The use of restriction fragment length polymorphisms (RFLPs) became a pioneering technique in DNA analysis.[6]

The breakthrough moment in DNA profiling came in 1984 when Sir Alec Jeffreys, a British geneticist, developed the first DNA profiling method at the University of Leicester.[4] Jeffreys utilized minisatellite DNA sequences, which exhibit high variability between individuals, creating a distinctive "fingerprint" for each person. This technique, known as DNA fingerprinting, was initially applied in criminal investigations.[4]

In the late 1980s, the invention of the polymerase chain reaction (PCR) by Kary Mullis revolutionized DNA analysis.[7]PCR allowed for the amplification of specific DNA segments, even from small or degraded samples, making it an essential tool for DNA profiling. This technological leap significantly increased the sensitivity and applicability of DNA analysis.[4]

During the 1990s, DNA profiling shifted from RFLPs to Short Tandem Repeats (STRs). STRs are shorter, more abundant, and exhibit high variability, offering enhanced discriminatory power. This transition facilitated the automation of DNA profiling processes and paved the way for the development of commercial DNA profiling kits.[4]

As the reliability and efficiency of DNA profiling techniques increased, many countries established national DNA databases to store and share DNA profiles for forensic and

investigative purposes. These databases have played a crucial role in solving crimes and identifying individuals.

In the 21st century, the emergence of next-generation sequencing (NGS) technologies has further transformed DNA profiling. NGS allows for the rapid and cost-effective sequencing of entire genomes, opening up new possibilities for comprehensive genetic analysis.

IV. PRESENT STATUS OF INDIA IN DNA PROFILING METHODS

The key techniques used in DNA profiling for legal purposes in India: [8]

1. Polymerase Chain Reaction (PCR):

PCR is a basic technique used in DNA profiling. It allows for the amplification of specific regions of DNA, making it possible to generate sufficient quantities of DNA for analysis, even from less or decomposed samples.

2. Short Tandem Repeat (STR) Analysis:

STR analysis is a widely used method in DNA profiling. Specific regions of the DNA, known as short tandem repeats, are amplified and analyzed. The number of repeats at each location is variable between individuals, providing a unique genetic profile.

3. Mitochondrial DNA (mtDNA) Analysis:

While nuclear DNA analysis is more common, mitochondrial DNA analysis is also employed in certain cases. Mitochondrial DNA is maternally inherited and can be useful in cases where nuclear DNA is degraded or insufficient for analysis.

4. Capillary Electrophoresis:

Capillary electrophoresis is used to separate and analyze DNA fragments based on their size. This technique is often employed after PCR amplification to analyze the amplified DNA fragments and determine the lengths of the STRs.

5. Automated DNA Sequencing:

Automated DNA sequencing techniques may be used for certain applications, particularly when detailed sequence information is required. This can be relevant in certain forensic and research scenarios.

(A) Future Possibilities

The field of DNA profiling is dynamic, and ongoing technological advancements promise to bring about transformative changes in the future. Here are some potential future possibilities and emerging technologies in DNA profiling:

1. Next-Generation Sequencing (NGS) for Whole Genome Sequencing:[9] [10]

NGS technologies continue to evolve, and the cost of whole genome sequencing is decreasing. In the future, widespread adoption of NGS for forensic DNA analysis may allow for comprehensive profiling of entire genomes, providing a more detailed and informative genetic picture.

2. Single-Molecule Sequencing: [11]

Single-molecule sequencing technologies aim to sequence individual DNA molecules directly without the need for amplification. This approach could overcome challenges associated with PCR amplification and provide high-resolution information for accurate DNA profiling.

3. Microfluidic Devices and Lab-on-a-Chip Technologies:[12]

Miniaturized lab-on-a-chip devices are being developed for DNA analysis. These technologies could streamline the DNA profiling process, reduce sample requirements, and enable more rapid and efficient analysis.

4. Massively Parallel Sequencing:[13]

High-throughput sequencing, here we can analyse multiple DNA samples. This could enhance the efficiency of forensic DNA analysis and contribute to the development of large-scale genetic databases.

5. DNA Methylation Profiling:[14]

DNA methylation, an epigenetic modification, can provide additional information about gene regulation and cellular identity. Future DNA profiling techniques may incorporate DNA methylation analysis for forensic applications and disease diagnostics.

6. Nanopore Sequencing:[15]

Nanopore sequencing involves passing DNA through a nanopore, and changes in electrical current are used to identify nucleotide sequences. This technology has the potential to simplify DNA sequencing processes and may become a valuable tool in forensic DNA analysis.

7. Artificial Intelligence (AI) and Machine Learning:[16]

AI and machine learning algorithms have the ability to analyse the data in DNA profiling. These technologies can aid in pattern recognition, interpretation of complex DNA profiles, and identification of relevant genetic markers associated with specific traits or conditions.

8. Rapid DNA Profiling Technologies:[17]

Advances in miniaturization and automation may lead to the development of portable and rapid DNA profiling devices. These devices could be deployed in the field for quick analysis, allowing for on-the-spot identification and investigative leads.

9. Enhanced Privacy Protection Measures:

With the growing concerns about privacy and data security, future developments in DNA profiling may focus on enhancing privacy protection measures. Techniques like differential privacy, which add noise to data to protect individual identities, may be explored.[18]

10. Customized DNA Panels for Targeted Analysis:[19]

Customized DNA panels targeting specific genetic markers relevant to forensic applications or disease susceptibility may become more prevalent. This targeted approach can enhance the efficiency of DNA profiling for specific purposes.

V. UTILITY OF DNA PROFILING [4] [20]

1. Criminal Investigations:

Example: A DNA sample is collected from a crime scene, such as blood, hair, or saliva. This can be used to find the identification through comparing it with the individuals in a DNA database (like CODIS), investigators may identify a suspect or link the crime to an existing offender profile.

2. Establishing Identity in Forensic Cases:

Example: In cases where the deceased is unknown, DNA profiling will help to establish identity by comparing DNA from the remains to DNA samples from potential relatives. This is particularly valuable in mass disaster scenarios or cases involving unidentified bodies.

3. Paternity Testing in Family Law Cases:

Example: In a legal dispute over paternity, DNA profiling of the child, the alleged father, and, if necessary, the mother, can definitively establish or exclude biological relationships. This information is crucial in determining child custody, visitation rights, and financial responsibilities.

4. Exoneration of the Innocent:

Example: A person convicted of a crime may request a DNA test to prove their innocence. If the DNA profile from the crime scene does not match the convicted individual's DNA, it can lead to their exoneration. Numerous cases worldwide have seen individuals released from prison after DNA testing demonstrated their innocence.

5. Cold Case Resolution:

Example: DNA profiling has been instrumental in solving cold cases that remained unsolved for years. A DNA match between evidence from a cold case and an offender's DNA profile, entered into the system later, can provide new leads and lead to the resolution of long-standing investigations.

6. Preventing Wrongful Convictions:

Example: In cases where there is doubt about a suspect's guilt, DNA testing can be crucial in preventing wrongful convictions. For instance, a DNA profile may reveal that the genetic material found at the crime scene does not match the accused, prompting further investigation.

7. Identification of Missing Persons:

Example: When a person goes missing, DNA profiling can be used to compare DNA samples from the missing person to profiles in databases. This process aids in locating and identifying missing individuals and reuniting them with their families.

VI. DNA GENEALOGY AND HELP IN LEGAL PROCEEDINGS [21]

DNA genealogy involves the use of genetic testing, typically through autosomal, Ychromosomal, or mitochondrial DNA analysis, to trace familial relationships and ancestral origins.

In the labyrinthine world of criminal investigations, the emergence of forensic genealogy has cast a new light on solving cases where traditional methods have hit a dead end. The Criminal Offender DNA Index System (CODIS), a comprehensive DNA database used in the pursuit of justice, has proven invaluable in linking crime scenes to known offenders. However, what happens when the DNA of a potential perpetrator is absent from this vast repository of genetic information? Enter the realm of forensic genealogy—a cutting-edge investigative approach that navigates the intricate terrain of familial relationships to unmask individuals eluding conventional detection. It is scientifically proven that "the apple doesn't fall from the tree" and "birds of feather flock together" and this is applied in forensic genealogy.

In scenarios where CODIS fails to yield a direct match, forensic genealogy steps into the forensic spotlight. By harnessing the power of genetic information derived from crime scenes and leveraging expansive genealogical databases, investigators embark on a journey through family trees, unravelling hidden connections and exposing latent truths. This method has emerged as a powerful tool in resurrecting cold cases, identifying unknown perpetrators, and

providing answers when the genetic fingerprint of a potential criminal remains elusive within CODIS.[22]

VII. ETHICAL IMPLICATION ABOUT DNA PROFILING

Some key ethical considerations related to DNA profiling:[24] [28]

1. Privacy Concerns: [23] [24]

Issue: DNA contains individual's genetic makeup, disease possibilities and also genealogy.

Ethical Implication: DNA profiling an database should respect individuals' right to privacy. Unauthorized access, data breaches, or misuse can lead to severe consequences.

2. Informed Consent:

Issue: Looking to the health illiteracy the consent provided by the person may not be good enough due to lack of knowledge about the impact of sharing it.

Ethical Implication: Ensuring that individuals are well-informed about the purpose, potential risks, and long-term implications of DNA profiling is essential to uphold the principle of informed consent.

3. Genetic Discrimination: [25]

Issue: Genetic information can be used for discriminatory purposes, such as in insurance, employment, or social contexts.

Ethical Implication: Legislation and policies should be in place to prevent genetic discrimination and protect individuals from adverse consequences based on their genetic profiles.

4. Family and Ancestry Discoveries: [26] [30]

Issue: DNA testing for genealogy purposes can unveil unexpected familial relationships or family secrets.

Ethical Implication: Individuals should be prepared for potential emotional and psychological impacts resulting from discoveries about their ancestry or familial connections.

5. Accuracy and Misuse: [27]

Issue: Inaccuracies in DNA testing can lead to false identifications or unjust legal consequences.

Ethical Implication: Ensuring the accuracy and reliability of DNA profiling methods is essential to prevent the misuse of genetic information in legal proceedings or other contexts.

6. Access to Genetic Information: [28]

Issue: Determining who has access to an individual's genetic information, including law enforcement, researchers, or corporations, raises ethical concerns.

Ethical Implication: Guidelines and regulations should define and restrict access to genetic data, ensuring that it is used for legitimate and ethical purposes.

7. Cultural Sensitivity: [29]

Issue: Cultural perspectives on DNA testing may vary, and certain communities may have reservations or concerns about genetic testing.

Ethical Implication: Respecting cultural diversity and engaging in culturally sensitive practices is crucial to build trust and ensure that DNA testing is conducted ethically.

8. Long-term Storage and Secondary Use:[23]

Issue: DNA samples and data may be stored for extended periods, and there is a risk of unauthorized secondary uses.

Ethical Implication: Establishing clear guidelines on the storage duration and purposes for which genetic information can be used is vital to prevent unanticipated consequences.

9. Consent for Familial Searches:[30]

Issue: In forensic investigations, familial searches may be conducted without the explicit consent of all family members.

Ethical Implication: Balancing the need for solving crimes with the right to privacy of family members requires careful consideration and ethical guidelines.

10. Ownership and Control: [31]

Issue: Determining who owns and controls the genetic information raises questions about individual autonomy.

Ethical Implication: Individuals should have a say in how their genetic information is used and shared, promoting autonomy and control over their own data.

VIII. CONCLUSION

In conclusion, the use of DNA profiling in legal proceedings has marked a transformative leap in the pursuit of justice within the Indian legal landscape. From solving heinous crimes to resolving complex familial disputes, the precision and reliability of DNA profiling have become integral to the fabric of the judicial system. As India advances towards the establishment of a National DNA Database, the potential for further breakthroughs in criminal investigations and the swift resolution of legal complexities is on the horizon.

However, as we applaud the scientific strides and the invaluable contributions of DNA profiling, it is crucial to tread the path of progress with ethical mindfulness. The ethical implications surrounding privacy, informed consent, and the potential for genetic discrimination demand our careful consideration. Striking the delicate balance between harnessing the power of genetic information for justice and safeguarding individual rights is imperative for the responsible and ethical use of DNA profiling.

As the legal landscape evolves, it is incumbent upon lawmakers, scientists, and society at large to foster a comprehensive understanding of the ethical considerations at play. Legislation and guidelines must be crafted to protect individuals from misuse, ensure transparency, and uphold the principles of consent and privacy. Moreover, cultural sensitivities and diverse perspectives on genetic information must be respected to foster trust in the legal system.

In this era where science and law converge, it is our collective responsibility to ensure that the use of DNA profiling not only resolves legal complexities but also aligns harmoniously with the ethical principles that form the bedrock of a just and humane society.

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