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Computational and Molecular Aspects of DNA Profiling in Forensic and Biomedical Sciences

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Abstract

DNA profiling has changed many diverse modern sectors, from health and criminal justice to ancestry research and animal conservation. It is one of the most important scientific instruments of our time. This technique makes it possible to identify people, analyze family relationships, and learn more about biology by looking at unique genetic markers in a person's DNA. DNA profiling has changed the way forensic science works by making it easier to find suspects, clearing innocent people, and making court cases more likely to end in a fair decision. DNA profiling has made progress in many areas besides forensics. It supports personalized medicine by allowing for therapies that are specific to a person's genetic makeup. It also helps with population genetics, evolutionary studies, and protecting biodiversity. Even while DNA profiling has many benefits, its broad usage creates ethical, legal, and privacy problems, especially when it comes to data security, permission, and possible misuse. This article discusses the scientific basis of DNA profiling, its many applications in the modern world, and the specific problems accompanying its rapid growth. It stresses the necessity for responsible governance to ensure that its benefits are realized while protecting the rights of people. DNA profiling operates on the premise that, although human DNA essentially is mostly the same across different groups of people, some parts of the genome are different enough to tell one person from another. Scientists can make genetic profiles that are statistically strong and very dependable by looking at these changeable regions. The improvement of lab techniques, along with better computer analysis and database administration, has made DNA profiling technology much faster, more accurate, and easier to use. DNA profiling has changed the way criminal investigations are done in forensic and judicial settings by giving investigators objective biological evidence that can connect suspects to crime sites or clear them of suspicion. Its use has made judicial systems around the world stronger by making it more likely that people will be found guilty and by facilitating reviews after a conviction. DNA profiling is very important for medical diagnosis, personalized treatment plans, ancestry testing, determining paternity, and protecting endangered species.

Keywords: Ethical issues, forensic science, genetic identification, modern biotechnology, personalized medicine

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INTRODUCTION

DNA profiling is the process of comparing biological evidence to see if a person's DNA matches that from a crime scene or another source. The procedure uses a unique DNA pattern that is found in each person's genetic code, which is like a fingerprint. DNA profiling is now a must-have tool for police around the world, and it has made it much easier to find the right people. A DNA match can be very strong proof in cases of paternity or ancestry. In numerous nations, forensic experts have also utilized DNA profiling to prove that people who were wrongfully convicted of crimes were not guilty. These applications illustrate the significance of DNA profiling for research, law, and society [1].

The main purpose of this paper is to talk about how DNA profiling works, how it helps society, and how it affects science and efforts to bring about justice in a bigger way. Some of the important words that will be explained include DNA, genetics, genetic markers, STRs, alleles, amplification, and the Combined DNA Index System (CODIS) [2]. This work addresses the inquiry: What constitutes DNA profiling? How does DNA profiling work? What are the main advantages of DNA profiling? What problems and restrictions still exist when it comes to DNA profiling? What are the legal and moral issues that are significant when it comes to DNA profiling? How is DNA profiling utilized in the real world? What does the future hold for DNA profiling?

WHAT IS DNA PROFILING?

DNA, or deoxyribonucleic acid, is the genetic material of all living things and many viruses. It carries the instructions for making proteins that are necessary for life. Two strands of nucleotides are twisted together in a double-helix shape to make DNA. A nucleotide has a sugar (deoxyribose), a phosphate group, and one of four nitrogenous bases: adenine (A), cytosine (C), guanine (G), or thymine (T). Genes are parts of DNA that tell cells how to make proteins. The arrangement of the nucleotides in a gene is what stores the information in that gene. The parts of DNA that are not coding are called “non-coding.” Many of these non-coding regions have different numbers of repeated sequences called short tandem repeats (STRs) or microsatellites. Forensic study finds these STRs very useful because they can still change while still providing a biological purpose. A DNA fingerprint consists of several STRs, each containing a distinct allele. Given that STRs are passed down through generations (Mendelian inheritance), a DNA fingerprint can be utilized to exclude persons or validate a familial connection. Countries, like the United States, have DNA databases for convicted criminals, people who have been arrested, and people who are missing. These databases can be searched for matches between DNA profiles and the attributes of crimes that have not been solved.

Profiling DNA means figuring out the DNA traits of a person to see if they are likely to be an unintentional contributor to a biological sample recovered at a crime scene. A DNA profile shows which alleles are present at a certain number of loci. It can be used for both personal identification and paternity testing. DNA sequencing and DNA profiling are like fingerprinting and photography in that sequencing gives a complete picture of all 3 billion bases in a person’s DNA, while profiling only captures a few loci that show naturally occurring differences between people. In many countries, the current forensic standard is to measure the STR profiles of 16 core loci, which are called the “STR set.” Each locus gives a genotype that is made up of two alleles. The number of loci is cut down to seven for real-time polymerase chain reaction (PCR). The presence or absence of specific alleles at certain loci in both mixed and single-source samples can yield additional relevant information. For instance, by interrogating just a subset of 12 autosomal loci coupled with the sex-determining SRY locus for male and female donors, it is possible to ascertain independently whether a sample is exclusively male, exclusively female, or a mixture of both [2].

HOW DNA PROFILING WORKS

DNA profiling uses samples from blood, skin, hair, saliva, and other biological materials. To avoid contamination, sample collection starts with careful observation and writing down what you see. The sample goes into a tube that keeps it safe while it is being moved and stored. In the lab, DNA is taken out, amplified by polymerase chain reaction (PCR), and then analyzed by capillary electrophoresis, which separates the amplified short tandem repeat (STR) fragments by size. After that, the electropherogram is read and a profile is produced. Different mixtures at a locus from different sources can make it harder to understand and alter statistical estimate. Mathematical models can help you figure out how strong the evidence is. For a correct interpretation and to show how important a match is, statistical probability is very important. Probability is a way of mathematically describing how uncertain something is. Probabilities go from 0 to 1: 0 means the event is impossible, 1 means the event is certain, and most events are given a likelihood in between. DNA profiles can be used to infer whether the profiles from two different samples come from the same source [2].

ADVANTAGES OF DNA PROFILING

Over the past few decades, the use of DNA profiling in criminal investigations has risen a lot. The principal objective of forensic DNA profiling is to generate biological evidence from bodily fluids found at crime scenes to identify suspects, convict offenders, or exonerate innocent individuals. The accumulation of this evidence forms the basis of centralized national DNA databases used for forensic DNA typing. The establishment and expansion of centralized national DNA databases have also contributed to improving crime detection and investigation on a global scale, with more than 69 countries operating such systems. When a profile from a crime scene matches someone in the database, the system gives investigators a straight lead. These national databases compare DNA profiles from crime sites with those of suspects, victims, or offenders. Testing multiple loci in autosomal short tandem repeat (STR) profiling reduces the chance of a coincidental match to less than one in a trillion, depending on the population under consideration [3].

PROBLEMS AND LIMITS

The first step in profiling involves collecting a sample from a biological substance such as blood, saliva, skin, hair, or other body parts. If stored correctly, the sample will stay safe for many decades after it is collected. The DNA extracted from it retains its profile-generating information far longer than fingerprints. Samples can even be procured from pasted stamps, envelopes, or bits of chewed food [4].

To extract DNA from a sample, the lab conducts several steps: first cleansing the sample of any debris and then dissolving it in a liquid that releases the cells' contents. Then, the fluid is heated to break down the cell membranes and free the DNA. After extraction, only little, easy-to-process pieces of the DNA from the sample are kept for profiling [5].

The fragments – a millionth of a millimeter long; invisible, many thousands in one hair – comprise those so-called short tandem repeats (STRs): the easiest genetic markings to detect. Each STR is made up of two, three, four, or five nucleotide sequences like GCTAGCTAGCTAGCTA. Thus, DNA profiling does not imply analyzing the entire sequence of a person's DNA.

The DNA that was chosen then goes through a pre-amplification process to make it easier for the lab to do the next selective amplification. For this, the lab needs full-size STR profiles that it keeps in a database. Profiles made before 1997, when the chosen DNA segments were defined, can still be relevant if the identical alleles are still in the profile database. Because DNA profiles on 1.0 and 1.1 systems can be easily reformatted, there is another reason not to keep specimens after profiling. Moreover, STRs can enhance supplementary information when a secondary selective amplification acts on the residual pre-amplified DNA.

THINGS TO THINK ABOUT LEGALLY AND MORALLY

DNA profiling is an incredible technology. It has changed criminal investigation. Massively. The public interest requires that legitimacy be both effective and open, and society benefits from a government and business sector that can find, confirm, or rule out persons as suspects or victims. Commercial forensic genealogy has added new features, but it has also pushed technologies that only a few people have seen. That nascent skill, that historical context, and that significant potential to aid law enforcement continue to provoke discussion. This kind of talk right now has an impact on the science policy around DNA profiling and on people's rights in business. F de Groot et al. [6]. talk about how important it is for everyone to have equal access to personal data and how decisions made today will affect future generations across borders. Very recent shifts indicate technology is set to enable whole genome analysis at lower cost and increased speed, with additional policy considerations becoming essential. These changes show that capabilities are growing quickly, and that civil society's promises to safeguard privacy, limit access, and own data will depend on open communication.

Confident laboratory capabilities and the right laws or rules are the basis for scientific and social effects. People should be able to grasp and trust the limits of guidance before they become permanent. DNA profiling supports wide ranging applications. It is still very important for people to trust both science and the government. Anything less makes it easier to be abused.

IN THE REAL WORLD

DNA analysis has helped police find suspects, make evidence stronger, and rule out innocent people in criminal cases. Analysis of instances from an earlier time has led to the overturning of wrongful convictions. Kirk Bloodsworth, Larry M. Johnson, and Keith Allen Harward are some of the most prominent cases of people who were wrongfully convicted of rape and murder and spent years on death row [1]. Medical uses include evaluating carriers and checking donor profiles in organ transplants. Tracing ancestry through DNA sequences has become a new service that combines genomic data with family history. Big forensic databases, like the Combined DNA Indexes System (CODIS), help by connecting cases that have similar evidentiary profiles [7]. DNA profiling has also helped find victims in disasters like the 2004 tsunami and the attacks on the World Trade Center [8].

THE FUTURE OF DNA PROFILING

The future of DNA profiling will bring about major technological improvements and problems with how it is used. Rapid developments in DNA sequencing technologies may help forensic DNA profiling. Next-generation sequencing platforms can quickly make a lot of sequence data. It will be possible to analyze the whole genome. Analysis will no longer be confined to a limited selection of loci selected for their forensic applicability. There are several efforts being made to investigate the legal, moral, and societal problems that next-generation sequencing brings up. People are quite worried about what whole genome sequencing could reveal about genetics and medicine if it were done at a crime lab. Policies are necessary to avert the dissemination of information beyond the designated forensic parameters. Recent studies look on how to provide fast DNA profiling while keeping medically related content private. Since the 1980s, the time it takes to analyze DNA has gone down a lot. Interest is emerging in creating a high-speed approach that doesn't require much lab or administrative work, making the capability available to everyone [9].

At the same time, new ethical and supervision problems come emerged, and possible ways to protect against misuse have not kept up with the apps. As DNA profiling becomes more common, strengthening monitoring and making decisions clear would help keep trust and responsibility in the process [10].

CONCLUSION

DNA profiling is good for society. It helps solve crimes, find people, answer issues about paternity and ancestry, help clear the names of those who were wrongly condemned, and provide researchers in many scientific field's direction. It is commonly accepted that it is an important part of current forensic science. Many uses show how powerful and flexible it is. Fixing the damage done by wrongful conviction has effects that go far beyond the case itself. DNA evidence can be quite convincing in crime scene investigation instances. DNA profiling helps with genealogical research, helps medical researchers learn more about how genes are spread in populations, and even helps answer questions about the relationships between ancient human remains and modern populations, which is important for understanding how modern humans migrated.

There are still big technical problems to solve, and more work needs to be done to make sure it has a future. Contamination of samples is an ongoing issue, and the possibility of gathering evidence from a compromised source heightens the risk of acquiring incomplete profiles or mixed samples. Inconsistent readings of statistics can unintentionally cast doubt on DNA tests that are almost perfect. Some people are right to worry that allowing genealogical queries to provide people access to extensive profiles could bring high-risk people to the public's attention. Talks in politics about the possible formation of a system for registering bodily materials also raise serious concerns. Lastly, DNA profiling is too expensive for underdeveloped countries and partners that want to work together [1, 2, 9].

DNA polymerase was the first enzyme to polymerize nucleotides. It was found in 1957. The free nucleotides that this enzyme uses as substrates were determined to be deoxyribonucleoside triphosphates, and they needed a single-stranded DNA template to turn into DNA.

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