

RECENT TRENDS IN DNA FINGERPRINTING

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ABSTRACT

DNA fingerprinting is emerging as an important tool for boosting criminal justice delivery system of the whole world. The technique has made tremendous progress in the recent past. The development of DNA database has been the focus of modern day research by compiling the population data of different populations. World's largest DNA databank, CODIS has made more than nine million DNA Profiling records. Different techniques and methodologies have been used by researchers in dealing with different types of forensic samples. Use of Polymerase Chain Reaction has revolutionized the field through its potential of detecting minute quantities of DNA samples. The use of quantitative PCR, a modified technique of PCR, has been of immense use for ascertaining the quantity of DNA in the samples and the extent of degradation apart from the assessment of the level of inhibition. All these advancements have made DNA profiling an important tool in boosting the crime investigating agencies and in turn the criminal justice delivery system in the whole world in general and India in particular.

Keywords:DNA Profiling, Polymerase Chain Reaction, Polymorphism, Restriction Fragment Length Polymorphism, Short Tandem Repeats.

INTRODUCTION

Establishment and evolution of forensic science, wherein the use of scientific aid is exploited in support of criminal justice delivery system, has been in use since times immemorial. The first written account of using entomology and medicine to solve criminal cases, is attributed to the book “washing away of wrongs” written by Song Ci in 1248 AD [1]. With the progress of time, more advanced methods for detecting the crimes were evolved in the fields of Chemistry, Physics, Document, Biology, etc. Upon adapting new ways of committing crimes to satisfy ulterior motives of criminals, there has been a progressive advancement in dealing with such crimes. One such technique, known as DNA fingerprinting or DNA profiling has gained a lot of scope in the recent past. The technique is unique in its power of discrimination between the individuals. It is based upon a molecule known as Deoxyribonucleic Acid (DNA) which is present in all the cells of an individual except non-nucleated RBC's. The DNA is present in the nucleus of a cell but can also be extracted from mitochondria and chloroplast (in plants). DNA is made up of millions of nucleotides which intern are composed of four basic subunits known as nitrogenous bases Viz. Adenine, Guanine, Thymine and Cytosine [A G T C]. The specific arrangement of these nitrogenous bases are responsible for coding different types of proteins. There are certain regions of the DNA which do not code for any protein and hence are known as non-coding regions. These non-coding regions are found to be highly repetitive. Sir Alec Jaffery in 1983while doing his routine work, observed

that there are certain regions of DNA which for being highly repetitive, are highly polymorphic in nature and can be used for identification at the individual level. This was the land mark development in the field of forensic science and in 1987 Robert Melias became the first person convicted through DNA evidence in UK [2,3]. Since its establishment, DNA fingerprinting has progressed by leaps and bounds in supporting the criminal justice delivery system. Development of DNA data base has greatly influenced the crime detection in countries like U.S and UK. CODIS (Combined DNA Index System) of United States, alone stores more than nine million profiles making it world’s largest DNA data base. Table 1, will give an idea about the progressive dependence of crime detection on DNA data base. “Forensic hit” refers to cases where match is made between two or more forensic profiles in the database whereas “Offender hits” refer to cases where an offender profile is matched to one or more forensic profiles in the database.

II.RESTRICTION FRAGMENT LENTH POLYMORPHISM (RFLP)

The first method used for DNA profiling was RFLP. In this technique the DNA molecule is subjected to restriction digest, wherein use of specific restriction endonucleases is made for cutting the DNA molecule in thousands of fragments of different sizes. The fragments so obtained are separated using the process of gel electrophoreses on the basis of fragment size. The smaller fragments shall move faster in the gel towards the opposite side of the electric field. These fragments can then be rendered visible through different methods, giving bands, sometimes also called bar code [4]. This technique has also made a great contribution in the Human Genome Project. This technique; however was not that much feasible for forensic samples. In most of the cases, in the field of forensic science, the samples carry DNA in degraded form and the sample size (DNA source) is very low that it becomes a limiting factor in RFLP technique which requires greater size of sample

Table 1. Number of Profiles in the NDIS, Investigations Aided, and Hits generated by Searches of NDIS

Year	Convicted Offender Profiles	Arrestee Profiles	Forensic Profiles	Investigations Aided	Forensic Hits	National Offender Hits (NDIS)	State Offender Hits (SDIS)	Total Offender Hits
2000	441181	-	21625	1573	507	26	705	731
2002	1247163	-	46177	6670	1832	638	4394	5032
2004	2038514	-	93956	21266	5056	1834	12482	14316
2006	3977435	54313	160582	45364	9493	4397	30138	34535
2008	6399200	140719	258943	81955	14364	8561	59184	67745
2010	8564705	668849	351951	130317	21983	15724	97772	113496
2012	9761083	1139065	436937	174680	28993	20698	132517	153215

Source: U.S. Department of justice, FBI, CODIS Brochure.

and very intact DNA.

III.POLYMERASE CHAIN REACTION (PCR)

Developed by Kary Mullis In 1983, PCR has revolutionized the field of forensic science. Although DNA is comparatively stable molecule due to inter-base hydrogen bonding, but storage of samples for too long and under non ambient conditions can definitely put a devastating effect on the yield of DNA. With the development of Polymerase Chain Reaction this problem has been solved to a large extent. Repeated cycles of fluctuating temperatures generate millions of copies of template DNA within a short span of time just like a Xerox machine. Care has to be taken with the quality of samples and its purification because PCR does not discriminate between the samples and is prone to inhibition by PCR inhibitors. Some common inhibitors are Viz. hematin, indigo, melanin, collagen, tannic acid, humic acid, calcium phosphate [5, 6].

IV.SHORT TANDEM REPEATS (STR's)

These repetitive sequences are highly polymorphic and hence are very useful in forensic DNA technology. STR's are divided in to different groups based on the length of repeats. However, tetranucleotide repeats are the most feasible repeats exploited in forensic DNA technology due to the fact that they produce lesser stutter products(due to slippage during polymerization) which creates ambiguity in further processing. Due to their bi allelic nature, STR's serve as important sources of polymorphism. Each individual receives one allele from father and one from mother at a particular marker and therefore will be highly useful in determining the identity of any individual by comparing it with any of the closely related individuals. Although the use of PCR in STR analysis has made it more powerful in forensics but the degradation of DNA samples poses a serious challenge in further analysis. In order to do away with this, the use of mini STR's has gained a lot of scope in the recent past. In this technique, the primers are bound closer to the markers and thus decreasing the size of amplicon. These reduced size amplicons are called mini STR's [7, 8]. But through this technique, the problem of low template DNA (LT DNA) and less stability of the markers used, cannot be ruled out exclusively. Therefore, to do away with this, the use of **Single Nucleotide Polymorphism** (SNP) analysis technique has gained a tremendous scope in the contemporary forensic technology. This has one more advantage over the use of STR's and that is it's capability of analysing highly degraded DNA samples. In this technique the occurrence of single base substitution, insertion or deletion is exploited for generating polymorphism. The low mutation rate in SNP's makes it highly stable and thus a reliable marker [7, 8, 9, 10]. Similarly, to combat with the problem of Low Template DNA (LT DNA), Whole Genome Amplification (WGA) is also growing in scope and importance. For example in one of its modified technique "Multiple Displacement Amplification" (MDA), the use of Phi 29 (Φ 29) DNA polymerase has a lot of advantage like higher fidelity and 3' \rightarrow 5' exonuclease activity.

The use of mitochondrial DNA found in mitochondria, also serves as an important tool for DNA fingerprinting. This source of DNA is different from the nuclear DNA and transmits through mother and hence has an

important role in studying maternal inheritance. Mt DNA has comparatively less discrimination power but is useful in highly degraded samples. The hyper variable (HV) regions are exploited for studying the polymorphism [7, 8, 11, 12, 13].

V.CONCLUSION

The field of DNA fingerprinting has a lot of scope of advancement particularly in India. Development of DNA database by making extensive and intensive population based studies will be a greater challenge in the country. Selection of desired and highly stable markers is the focus of present research and different reputed companies are developing more reliable kits to meet out all the requirements in the field of forensic science. New approaches will be explored to make the technique more and more error free.

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