

ROLE OF BIOINFORMATICS IN FORENSIC SCIENCE

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خلاصہ

’جیاتنی علم الاطلاعات‘ ایک ایسی انجینئرنگی ہوتی ہے جو جدید طاقت ہے جس نے سائنس کے تمام شعبوں بشمول علم قانون طب، کو اپنے زیر سرایہ کیا ہوا ہے۔ ہرگز رستے وقت کے ساتھ ساتھ جیاتنی علم الاطلاعات کی دنیا دیر بنائے جانے والے آلات کی پیداوار، آفاقیات اور فضیلت علم قانون طب کے حوالے سے اہم ہوتی جا رہی ہے۔ علم قانون طب کا چھوٹے سے چھوٹے عمل جیسے کہ کسی معمولی جراثیم سے ڈی این اے کے ٹکڑے کا عمل ہو یا ڈی این اے کی نقشہ جاتی، جنسی شناخت ہو یا اعلیٰ درجے کی خود ساختہ شخص شناخت، اقرا می نظام ہو یا کمپیوٹر کے اصولوں پر مبنی آلات کا استعمال، سب کامنکات میں شمار ہو چکا ہے۔ مزید یہ کہ این جی ایس ٹیکنالوجی اور تو لیدیات، ایک ہی وقت میں ایک سے زیادہ خلقی مقام کے بہت سے مختلف جینیاتی مفادات کا تجزیہ کرنے کی صلاحیت رکھتی ہے۔ اس مزید نے وسیع پیمانے پر تحقیق کے دروازے کھول دیئے ہیں۔ یہ پیمانہ و امان کے لحاظ سے کی گئی کمپیوٹر ازجگ تحقیق اور ان کے محرکات اور فوائد کے بارے میں لکھا گیا ہے۔ ظاہری شقیوں کے مجموعی مدد سے تمام کمپیوٹر ازجگ طریقوں کا ایک تنقیدی جائزہ لے کر ان کے نقصانات و مفادات اور ان کی افادیت کا اندازہ لگایا گیا ہے۔ ان طریقوں میں سے ڈی این اے کی نقشہ جاتی، ایس ٹی آر نقشہ جاتی، اے ایف ایس آر ایلو گرام ہر تہیات، ڈی این اے کے شقیوں کی ذخیرہ اندوزی اور این جی ایس وغیرہ شامل ہیں۔

Abstract

Bioinformatics, an emerging powerful science gradually enfolds all sorts of sciences under its huge banner; Forensic Sciences being one of them. With the passing time the productiveness, efficacy and virtue of Bioinformatics based tools is taking over the Forensic Sciences and research. Whether it be declaration via extracting the tiniest DNA from the tiniest microbial sample, DNA profiling, gender identification or the most advanced Automatic Fingerprint Recognition System, computational tools are aiding the Forensics in every possible way. Furthermore, Next Generation Sequencing technology along with the epigenetic studies gives rise to a vast research domain by simultaneously analyzing multiple loci that are of forensic interest in multiple genetic contexts. The paper evaluates then boosting influence of computation on this immensely significant field, especially in terms of law and order. A series of physical evidences are put up for the critical analysis that reveal pros and cons and definitely the efficacy of the techniques that are being implemented including DNA profiles, STR profiles, AFSR algorithms, sequencing, computer review, storing DNA evidence, Next Generation Sequencing and others.

Introduction

Bioinformatics is basically an emerging field that marks the union of computer and biology. A discipline where applications of computer science are applied to biological data with an aim to examine data in various possible ways like Sequence analysis, image analysis, sample management and population modeling. Mentioned are some of the methods that come under the vast field of Bioinformatics. Through these techniques, it becomes possible to process and understand the bulk biological data. Molecular biology, computer science, and statistics are three distinctive fields, which contribute in the assembly of tools and techniques applied in Bioinformatics. With time, the implementation, evolution and usefulness of the computational techniques is being enhanced, some of them include DNA amplification with the help of PCR, by using prediction method exact boundaries of genes are calculated, for DNA mapping, to analyze phylogenetic reconstruction and one of the trendiest use of bioinformatics is in the field of forensic science Ahmed *et al.*, (2014).

Ajay *et al.*, (2012) defined forensic science as a blend of criminal integrity and science. Forensic Science is the scientific innovation that is valuable in inquisition of crime and the information that is obtained from the crime scene is scrutinized from all aspects. Finally, this data is used to investigate the related transgression, then these facts are put forward in the court that's quite helpful in order to castigate the criminal. These days' bioinformatics is widely acceptable in the field of forensic science because, with the help of computational tools, it became quite uncomplicated and reliable to gather evidence regarding a particular crime scene.

Forensics requires bioinformatics

Currently, in order to have forensic assessment and evidence such experiments are designed by Redon *et al.*, (2006) which are combination of bioinformatics and forensic statistics. Bioinformatics has a great impact in

making hypothesis on DNA samples with the help of forensic statistics. In DNA sampling, while examining population of identical units, decision is made on smallest population sample size and test results of statistical importance are also influenced by bioinformatics. States in microbial forensics bioinformatics plays its part by recognizing the microbial samples in a very short amount of time that contains multiple levels of declaration. In order to attain desirable results, methods are supported by assessment of superior genomic sequences and precise determination which are helpful to identify protein structure, gene composition and also useful association between dispersion and virulent systems Slezak *et al.*, (2005).

DNA testing

According to Forensic bioinformatics basic task is to make advancement in setting up the forensic records which is useful to store rough draft of DNA of criminals that is taken from the crime scene and later presented for DNA testing (Ajay *et al.*, 2012). Statistical and technological progressions i.e. learning algorithm based on machine learning, DNA microarray sequencing, and thin film transistor biosensors etc. are used to improve accuracy and authenticity of the results. Now a days genetic tests have been extensively used for detection of mass fatality and forensic evidence as well. A multidisciplinary panel including medical examiners, fingerprint professionals and forensic pathologists gather the data which is then incorporated with the results of genetic testing.

Person's Identification

Most frequent proofs of suspect are described by Mahajan *et al.*, (2015) which include alcoholic drink bottles, hair, and cigarette butts. It is such a tough task to identify criminal on the bases of such kind of evidence because forensic investigation demands DNA extractions and there is more probability to have impure DNA from such kind of material that would be problematic for further investigation. In criminal analysis, Alenazy *et al.*, (2015) revealed that the bloodstains proved to be foremost corporeal proof. According to Nandan *et al.*, (2015) any individual can be identified with respect of physiological characters i.e. the lip print pattern is a unique among individuals and thus helps in the identification during the forensic investigation, Subramanian and Jagannathan (2015) states that fingerprints, hand palms, biological typeset i.e. saliva, blood and behavioral distinctiveness which includes lip motion, dynamic signature, dental analysis etc. are helpful in forensics and according to Manjunath *et al.*, (2011) good quality DNA can be obtained from suspect's teeth. Human beings have 99.9% DNA identical and only 0.01% is enough to make difference among human race as with respect to different environmental factors, each person responds differently (Redon *et al.*, 2006). The foremost subject matters of forensic DNA analysis described by Huffine *et al.*, (2001) and Biesecker *et al.*, (2005) are the individual recognition and association with other persons.

Forensic investigation usually has to deal with kinship quarrels like parenthood disputes, recognition of any dead body and state of suspected incest are included in most cases. It also deals with the frauds related to nutrition like toxic food, recognition of semen for unfaithful suspect, criminal issues are addressed in forensics on priority bases. In case of severe accident, postmortem report is required for individual recognition that is essential for accident analysis as well. In case of any natural disaster or sudden mishap like terrorism or plane crash, genetic tests are carried out extensively.

Latest work in the field of forensics is the preservation of DNA for extended period and sampling of large amount of tissue that is possible in the dehydrated condition along with the applications in mass fatalities. Bianchi and Liò (2007) states that there is intense repetition occur in the genome of humans. DNA sequence length scale, distribution and their number are the sites where recurrence takes place like intermingled families, hundred base pair lengthy transportable elements etc. In human genome, there are more than a million *Arthrobacter luteus* (ALU) sequences which are about 300bp in length and they have ability to make their replica in order to produce mutation. In forensic DNA typing, such methods are essential that help in identification of genetic discrepancy among humans, variable number of tandem repeats being used for the purpose.

Gender Identification

According to Delgado *et al.*, (2005) and Mitchell *et al.*, (2006) bioinformatics can be used to take full advantage for forensic analysis. For gender identification, amelogenin marker testing is required. Amelogenin being a protein that is made up of about 90% of tooth enamel protein. About 106bp are obtained from X chromosomes and 112bp from Y chromosomes with the use of primer that is specified for intron 1 of the amelogenin gene. Results illustrating that two bands are shown by male source (XY) and one band is shown by female (XX) on an agarose gel. Petricevic *et al.*, (2006) states, an individual can also be identified by tracing DNA on bedding.

Parentage testing

Forensic bioinformatics play crucial role in child parental issues. Prasad (2008) illustrated a case about a Ghanaian youngster, his birthplace was UK but didn't allowed to go back to UK during his visit of Ghana. Though his family was in UK so in order to confirm boy's pedigree the lawyer suggested for DNA investigation. Immigration agency quit the case as the test report revealed that the woman in UK was the true mother of the boy, so he was allowed to go back and joined up his family.

STR system

Alenazy *et al.*, (2015) states that one of the main physical facts in criminal examination includes blood stains. It is observed that in most of the crime scenes, very diminutive blood stain is obtained; the reason is that the executor tried his level best to remove all indication that may become the source to disclose the criminal's identity. Short tandem repeat (STR) identification method is useful in such incidents by taking that minute blood sample on which probability test had been done on prior bases. STR technique is very advantageous because this technique can be applied on huge range of material which includes cigarette edges, criminal's tissues on bullets and on gun stifle, mummified infant, dentinal stuff, dried bubble gum, dead bodies destroyed by fire, in strangulation case; epithelial cells of criminal obtained from the neck of the sufferer, human feces or urine, decomposed body elements, skeleton remnants, body parts after massive destruction etc Bianchi and Liò (2015). These days STR system has incorporated in DNA identification technique for criminal analysis. Biological facts are very valuable in order to identify association between sample and real criminal action (Ahmed *et al.*, 2013).

Microsatellites

Microsatellites hold immense importance in forensic DNA analysis. These are present in genomes of all living organisms and these tandem repeats are found in the form of preserved tiny fragments of DNA and there are different types of tandem repeats which include variable number tandem repeats (VNTRs), simple sequence repeats (SSRs) and short tandem repeats (STRs). The main reason to use microsatellite is that it has high polymorphism marking it as an influential genetic marker to facilitate mapping of genome of numerous organisms. Muniruzzam *et al.*, (2015) in human DNA, microsatellite loci can be identified through hybridization technique that is comprised of major sequences' tandem repeats. Southern blotting hybridization gives definite details of suspect and produce such kind of finger prints that have numerous variations and also the consistency of germ cell as well as somatic cell has been demonstrated with this technique Gill *et al.*, (1987). Polymerase Chain Reaction (PCR) that is used for amplification purpose for a particular copy or small number of copies of DNA, through this technique a huge number of copies can be produced. In present scenario forensic science and in paternity as well PCR is used to identify genetic fingerprints Zubakov *et al.*, (2010).

AFRS algorithm

Jeyanthi *et al.*, (2015) states that in crime cases, fingerprints of the covert are obtained to precede investigation as this information play significant role in identification of criminal but at times even very little amount of physical interference makes the fingerprint image partly covered and to facilitate fingerprint recognition it's very important that the image is capable to split. For this purpose, an algorithm is developed that is called Automatic Fingerprint Recognition System (AFRS). This algorithm is designed in such a way that can eliminate any overlies dormant image from the required image and provide clear result of the given image.

Next generation sequencing and forensics:

Next-generation sequencing (NGS) technology due to its high-throughput capacity and extremely low cost, has emerged as a very essential analytical tool for almost all genomic researches and the fields related to them including the Forensic Sciences Weber-Lehmann *et al.*, (2014). The incorporation of NGS in Forensics harnesses several domains of research that include the parallel analysis of loci of forensic interests in autosomal mitochondrial, sex chromosomal and almost every possible genetic context.

Yang *et al.*, (2014) illustrates what actually is NGS, it refers to non-Sanger based high-throughput DNA sequencing technology. Simultaneous sequencing of millions or billions of DNA molecules, minimizing the need for the fragment based cloning method used by Sanger sequencing. This generation of enormous number of reads paved the way of NGS to all sorts of fields including Forensics. Moreover, NGS technology holds its significance in others aspects of research. Precisely mentioning as DNA database construction, phenotypic inference, ancestry, body fluid and species identification via it, monozygotic twin analysis, and may also help proceed forensic animal and plant detailed microbial analysis Weber-Lehmann *et al.*, (2014).

The DNA technologies holding application in forensic investigations has provided DNA analysis an important role in forensic science. Unlike numerous other fields of science, the forensics has to somehow confront with the data template of extremely low copy number, highly-degraded, raw and full of contamination. So, high efficacy and accuracy along with time and cost considerations are ultimately the major needs. The capillary electrophoresis (CE)-based fragment analysis of Sanger method comes with its limitations of inability

to analyze the multiple genetic polymorphism in a single go or a reaction, leading to low resolution and loss of useful genomic information Rizzi *et al.*, (2012). But this limitation highlights the need to explore the usefulness of NGS and it being the need of the hour.

Epigenetics

A lot of evidences and studies ultimately declare the DNA sequencing analysis a powerful influential tool in forensic identification Jeffreys *et al.*, (1985). Thus, epigenetic markers being essential building block for the purpose has various applications in forensic science. One major application of epigenetic markers with evidence is their distinguishing capability of monozygotic twins Li *et al.*, (2013) and precisely determining the age of the DNA donor. Some of the epigenetic approaches which make the NGS technology their base are whole-genome bisulfite sequencing, methylation bead chips, reduced representation bisulphite sequencing and the list continues Bocklandt *et al.*, (2011). All of the mentioned sequencing methods hold a requirement of large amount of DNA, making it crucial for the forensic epigenetic analysis to occur. In contrast, extremely sparse amounts of starting DNA can trigger the analysis through genome-wide amplification of a bisulphite-modified DNA. Also, bisulphite genome DNA sequencing with the micro-volume blood spot samples also occurs. Xu *et al.*, (2012). Weber-Lehmann *et al.*, (2014) described how identification of extremely-rare mutations by ultra-deep NGS can differentiate between MZ twins, providing a solution to paternity and forensic cases involving MZ twins.

Discussion

Bioinformatics tools are very helpful in forensics but there is still need to be more careful while generating results from computational tools because at times there is discrepancy arise between set of statistical rules and biological reaction. As the most doubtful results produced in phylogeny reconstructions and ClustalW reconstructed alignment. Ahmed *et al.*, (2014) It is also observed that correct alignments are generated from those sequences which are very closely related with the help of bootstrap method. At the same time, it is expected that the alignments which are produced from biological sequence sets produced inaccuracy in more than half of the alignments so such method is used to determine the constancy of tree topology but not give accurate phylogenetic tree. But with the passage of time, there is an improvement in results and the computational programs are becoming more consistent progressively. Parentage testing and family reunification is also something that comes under category of bioinformatics and forensics. Though it's very useful but many people condemn such test as it interrupts their privacy Weiss (2011). In last 20 years, the field of bioinformatics has become more advanced and the objective of production, as well as assemblage of various documentation and investigative tools, has been accomplished. Worldwide, public realm assets such as GenBank have become very crucial source for research purpose. Prasad (2008) currently, lives of millions of people globally influenced by the forensic DNA technology. This approach is still getting high rate of approval on universal level. Forensics played well in major events like in 9/11 activist assault, the victims were recognized through DNA profiles analysis. Biesecker *et al.*, (2005).

Nowadays Forensic DNA databases fast expansion put many questions on the standard of data related to placing and its maintenance, uncertainties related to its effectiveness and there are also chances of confidentiality violation of such huge private data Ge *et al.*, (2014). On the other hand, in earlier period various types of transgressions put under DNA investigation and as a result numerous DNA profiles produced which become helpful to generate novel measures i.e. in Familial DNA Database Searching, find similarity between DNA profile of executor's family member and the data collected from crime scene and the first victorious familial search was carried out in 2004 in UK that confirm Craig Harman is responsible of assassination but many countries are against to use this type of facts i.e. according to Germany viewpoint, it is important for each autonomous society to enjoy freedom and constitutional rights that's why expansion of forensic database is discouraged Wallace *et al.*, (2014).

In Pakistan DNA profiling has many imperfections. There is deficiency of funds, professionals, and data protection and also, there is insufficient guidance as well as improper apparatus. There is need to give more advance training to Pakistani analyst. Weiss (2011) In Pakistan, there are some institutes i.e. University of Veterinary and Animal Sciences Lahore (UVAS), Government College University Lahore (GCU) and University of Punjab (PU) initiated DNA forensics research. Center of Excellence in Molecular Biology (CEMB) is a committed laboratory started in 2005 and it deals with cases include crimes, catastrophes, and paternity clashes. Higher Education Commission (HEC) should focus on this field also by acquiring advance strategy in association with law enforcement institute facilitates the forensics Farooq and Waheed (2013).

Conclusion

Finally, it is concluded that Forensic Investigation is impartial valuable computation for DNA recognition with the help of bioinformatics. Complete features of DNA profile of an individual can be obtained through this analysis Hedman *et al.*, (2010). According to Levitt *et al.*, (2007) though there are many believers who think

that in future DNA will be the solution of every related problem but one should not be fully dependent on forensic DNA profile because there is no certainty that this technique will overrule on expenses related to societal and moral values. There is need to introduce more authentic techniques. This practice is on the way of advancement and will keep pace with the modern world in very short duration.

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