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REVIEW ARTICLE

RECENT TECHNIQUES BASED ON THE UTILIZATION OF DNA AND AUTOSOMAL SINGLE NUCLEOTIDE POLYMORPHISMS FOR IDENTIFYING HUMANS

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ABSTRACT

The biological samples used in forensics can contain DNA which is highly fragmented as a consequence of exposure to any of the numerous degrading factors. Analysis of the sequence or size of the products of Polymerase chain reaction is at present responsible for the analysis of remains of humans in forensics. Despite the effectiveness of protocols based on PCR, there are certain limitations that are presented by the low numbers of copies of the template and the variations that are imposed by the decaying process to the template. The primary aim of this research is to explore the significance of autosomal Single Nucleotide Polymorphisms (SNPs) in forensic science through the identification of humans at a crime scene. This study provides an exploration of the applicability of autosomal SNPs for the identification of humans at crime scene. This would fill the gap present in the current literature regarding the significance of autosomal SNPs in the identification of humans during crime scene investigation. It will also enable the identification of the criminals involved in several types of the crimes ranging from general theft to rape and sexual assault, murder, and robberies. It will also allow the identification of dead bodies in cases where it is difficult to identify the dead person due to unrecognizable condition of the body. This study will facilitate the improvement of the investigation of crime scene investigators. It will provide a significant way for the incorporation of recent techniques of the molecular genetics into forensics. Reduction in the workload of the crime scene investigators would also occur through the implementation of outcomes of this study into the field of forensic science. There are several studies which have demonstrated the applicability of SNPs in forensic investigations for identifying the humans at crime scene. Several effective and efficient technological systems have been developed by the researchers which are capable of performing analysis of biological samples containing degraded DNA because SNPs can be obtained from these samples. Physical characteristics of the individuals can be predicted through the analysis of SNPs. This can provide significant information about the color of eye, hair and skin of the individuals involved in crime.

KEY WORDS: Single Nucleotide Polymorphism; DNA; Polymerase chain reaction; Forensic; Molecular genetics; Autosomal; Chromosome; Genes; Alleles; DNA Sequencing.

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INTRODUCTION

Utilization of DNA for the identification of human beings has become an important aspect of the application of molecular genetics in forensics. DNA has been utilized for the identification of criminals involved in various criminal activities.[1,2] Once the DNA of the criminal has been collected from the crime scene, the identification of the criminal can be performed through analysis of the sequences of the DNA with that of the records present in the national database of the concerned country.[3] In most of the crimes, it is difficult to deliver justice since the identification of criminals is complicated. However, the investigators can overcome this hurdle if evidences are available at the crime scene as biological material like blood, bone, hairs, teeth, semen and saliva. Extraction of the DNA can be performed from these

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biological samples even if only a single cell of the criminal could be extracted for analysis.[4] Identification of the criminals through finger print matching has been performed extensively but this technique has not provided significant results as the criminals have developed the strategies which have made it impossible to collect the finger prints from the crime scene.[5] Use of gloves and coating of skin with other means to hide the finger prints has become a common practice of the criminals. This has developed the need for the identification and application of a technique which could provide significant benefits regarding the identification of the criminals at the crime scene.

Sometimes it is also difficult for the investigators to recognize the dead bodies at the crime scene, in cases where the victim has been brutally murdered by the criminal. This develops a critical situation not only for solving the crime, as the identification of the criminal cannot be performed but also because it becomes difficult to identify the victim if there is no evidence for that crime. This has resulted in the development of need of application of molecular techniques in forensics to enable the identification of the victim also. At present, researchers have focused on the use of Autosomal Single Nucleotide Polymorphisms (SNPs) for the identification of humans. This requires the exploration of this specific aspect of molecular genetics for utilization in forensics to enable successful investigation through the identification of the criminal and victim.

Topic choice Rationale

There are several reasons that have motivated the researcher for choosing this particular topic for performing this study. The major reason is the increased prevalence of crime rate in the world. According to the statistics of the survey of Bureau of Justice Statistics in USA, there has been a rise in the rate of violent crimes including robbery, sexual assault, forceful, and simple assault. This has resulted in an increase from 22.6 victimizations per 1,000 persons in the year 2011 to the victimizations at a rate of 26.1 in the year 2012. There has also been an increase in the rate of property crime including household burglary, theft of the motor vehicles, and general theft. This resulted in the increase from 138.7 victimizations per 1,000 households in the year 2011 to 155.8 victimizations in the same study.[7] Secondly, numerous advancements have been made in molecular genetics which have paved the way for the application of the techniques of molecular genetics in various fields. Consideration of the success of various techniques of molecular genetics in forensic science has prompted the search of the applicability of autosomal SNPs for identification of humans at crime scene. It has been presumed that it would aid the forensic investigation in the identification of criminals so that the rate of crime might diminish in the any country like Pakistan.

Aim and Objectives of the Research

The primary aim of this research was to explore the significance of autosomal SNPs in forensic science through the identification of humans at crime scene. The objectives of this study were as follows:

1. To review the literature that demonstrates the usefulness of autosomal SNPs in forensic science.
2. To suggest the application of autosomal SNPs in forensic science based on the outcomes of the review.

LITERATURE REVIEW

DNA is responsible for the formation of genes which are the heredity units containing the instructions for the formation of proteins. There are usually two copies of each gene in the eukaryotic organisms, each one present on one pair of the chromosome. The inheritance of these genes is based on the transfer of one gene from the father and one from the mother. The presence of alleles which are the varying forms of the same gene is responsible for imparting the various characteristics to the organism.[8]

Application of DNA in Forensics

DNA has been used extensively in forensics for the identification of victims through the assessment of the biological relationship and individual identification. This has become possible due to the efficiency of DNA technologies and their accuracy for performing the identification of the criminals and victims. Databases of the forensic DNA and forensic DNA technology have been recognized widely as the most effective and efficient tools available to the organizations of law enforcement for fighting crime. The application of a combination of computer science and biological science has provided the DNA evidences strength to be utilized for conviction and identification of the guilty and exoneration of the innocent.[9]

Recent Techniques based on the Utilization of DNA for use in Forensic Sciences

There are several techniques that have been utilized by the researchers and investigators for the identification of DNA of an individual leading to the identification of the person. These techniques are based on the modern methods which have been implied in the scientific research for the identification purpose based on the application of technological and biological aspects. These techniques include numerous processes such as DNA fingerprinting, polymerase chain reaction, and DNA sequencing for applying the techniques of molecular genetics in forensic science.

DNA Fingerprinting

The process of DNA fingerprinting is based on the collection of a sample of cells which contains DNA.

These samples include blood, hair, and skin which can be used for the extraction of DNA followed by its purification. Later on, the strand of the DNA is cut at specific sites with the help of restriction enzyme resulting in the formation of strands of various lengths.

Autosomal Single Nucleotide Polymorphisms (SNPs)

Autosomal single nucleotide polymorphisms are the most common type of the genetic polymorphisms which have alleles that are associated with particular physical characteristics and specific populations. Polymorphic genetic markers of the proteins have been used during the past twenty five years for performing the differentiation of the individuals. There were several factors that limited the utilization of protein based genetic systems. The basis for the inapplicability of these systems was dependent on their inability to perform individualization of the individuals.[10] These limitations have been overcome by the use of genetic typing of the genetic polymorphisms at the level of DNA. These techniques enable the forensic scientists to exclude those individuals who have been related falsely with the biological samples obtained from the crime scene. It also reduces the number of the individuals who are considered to be related with the biological sample. The initial system that has utilized the PCR based genetic mapping techniques was based on the variation of the single nucleotide polymorphisms. There are some biological samples in the forensics which cannot be analyzed. These biological samples are generally inadequate in amount containing small or insufficient amount of the DNA or are degraded.[11]

Application of SNPs in Forensic for Identification of Humans

The only applicability of SNPs in forensics is in the cases where missing people are required to be identified. In such cases, the ante-mortem samples collected from the victims or from the relatives of the victims are subjected to forensic analysis. These samples are collected denovo followed by the genotyping of the samples with the remains of the victim. This has resulted in the replacement of the Standard Terminal Automation Replacement System (STARS) with Short Tandem Repeats (STRs) since the DNA which is available in these cases is highly degraded. The application of SNP markers have been identified in several cases for Doppler Velocity Index (DVI), which also included the terrorist attacks at the world trade center.[12]

Identification of Victims

It has been expected that the application of identification of biogeographic ancestry of an individual based on the DNA dependent processes can be useful for the identification of victims who are unrecognizable due to brutal murdering and various other reasons. This will also find application in the identification of the victims of terrorist attacks, as it is difficult to identify the victims of such cases. The period of the applicability of biogeographical ancestry of the individuals in the forensic investigation is difficult to predict. It could be determined at the point of detection of the geographic substructure through the utilization of sets of DNA markers. The knowledge present at the current level provides information about the large geographic regions including continents, although differentiation at the level of sub-regions can also be performed. The restriction of the genetic diversity is not followed by the political borders.[13]

Synthesis Methods

The data for this study was provided by the review of the literature which included all the studies that were relevant from data base. The interpretation of the observations of the literature review was performed through an analysis of the collected information. Links between the various parts of the analyzed data were formed through the synthesis of the collected ideas. Synthesis also presents the mode in which the researcher presents the conclusion of the collected material from the literature based on the research question. It is essential to perform the procedure of synthesis in a gradual manner so that the gathered data can become convenient to understand.[16]

Data Collection

Online literature search was carried out to collect relevant information through different search engines like Google and Google Scholar and databases like PubMed, PubMed Central, Cochrane Reviews, ScienceDirect and research repositories of various universities. The search was limited to the publications during the period from 2006 to 2016 in English language only.

A thematic analysis of the studies is presented to develop the understanding of various aspects of SNPs application in forensic investigation for identification of humans at crime scene.

DISCUSSION

SNPs are the newly explored markers that are being explored for their ability to predict the biogeographical ancestry of the humans. They have also found application in forensic genetics as they can be used for identification of the humans at crime scene, whether the individual is a victim or a criminal. SNPs are also able to be analyzed and extracted from the biological samples of the crime scene which are degraded due to various environmental factors or contain DNA in very low amount.[17] They have been successfully employed in the analysis of biological samples of teeth, bone, hair, saliva, blood, and sperm which were nearly 1400 years old. The ability of SNPs to be used from the samples which have been exposed to environmental factors resulting in their degradability is the basis for their suitability to be applied

in forensic identification of humans.[18,19] SNPs can be used for the prediction of paternal and maternal ancestry because Y-SNPs and mitochondrial SNPs can be analyzed for suggesting the biogeographical ancestry of the individual.

Several systems have been designed that can be used for analysis of SNPs so that they can be used for the identification of humans at crime scene. GenPlexTM HID System has proven to be significant for performing analysis of SNPs. This system can be used for the identification of various geographical populations as it was able to analyze SNPs of both the European and the US population.[20] Typing of all the SNPs can be performed by alteration of buffer of another system which suggested its efficiency for application in forensic genetics to identify the humans at crime scene.[21] SNPs are important for the identification of individuals because they can predict the phenotypic characteristics of the population. These characteristics range from the color of eye to color of hair and skin along with the prediction of age and height of the individual. It has been observed that SNPs provide considerable amount of data regarding the color of eye which can be used for the identification of individuals. Color of skin can also be determined through the application of SNPs in forensic identification of criminals.[6]

CONCLUSION

Autosomal SNPs are the mutations at a specific point in the DNA. They can be extracted from the biological samples of the DNA that are highly degraded. They have been used previously for the identification of biogeographical ancestry of the individuals belonging to diverse geographic regions. This has lead to the evidence for the applicability of SNPs in forensic analysis for identification of the criminals and victims through the prediction of their hair, eye, and skin color. Height and age of the humans at crime scene can be determined by the application of SNPs in forensic analysis.

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CONFLICT OF INTEREST
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