

**Title: Genetic identification : A Review On Autosomal Short Nucleotide Polymorphism's as diagnostic Tool for Identifying Human.**

**Running Head : SNP's for Identify Humans**

### **Abstract**

Advancement in the field of genetics has resulted in the application of several techniques of molecular genetics in forensics. The body of human beings is composed of DNA which is a chemical molecule responsible for imparting phenotypic and genotypic characteristics to the individuals. The most recent advancement of molecular genetics, which has found application in forensic science, is the use of autosomal SNPs because they can provide information about the ancestral genetics of human beings. The primary aim of this research is to explore the significance of autosomal SNPs in forensic science through the identification of humans at a crime scene.

A secondary qualitative research design has been selected for conducting this study. This secondary research is based on a systematic review of the studies which have provided an insight in the significance of autosomal SNPs in forensic sciences by using various Data search Engine . Studies that were not performed in English language or performed before 2010 were not included in the review. SNPs can be used in the forensic investigation for the identification of individuals present at the crime scene.

**Key words**

(SNPs) Single Nucleotide Polymorphism , Deoxyribonucleic acid (DNA) Short tandem repeats (STRs), Biogeographic Ancestry, signal joint T cell receptor excision circles (sjTRECs

## **Introduction**

Recent advancements in the field of molecular genetics have contributed to the development of techniques that have provided benefits to various other fields. Molecular genetics deals with the analysis of the genome of organisms at molecular level. There are numerous genes that constitute a genome of the humans. There are approximately 3 billion base pairs in the human genome (Kwon, 2013). These genes play a crucial role in the development of the DNA which is the core element for the formation of human body. Each cell of the body of an individual contains the DNA which is a chemical code. There is a similarity in nearly 99.9% of the sequences of DNA of humans in every individual. Therefore, the remaining 0.1% of the DNA sequences that are different forms the basis of the differentiation of an individual from the other (Pongracz, 2013). This variation enables the identification of a person and imparts the different characters to the human beings that vary from person to person.

Sequencing of the human gene has been performed over the past few years which has enabled the scientists to analyze the sequence of genes in the DNA. It has served as the foundation of research in the modern biomedical research. The technology of DNA sequencing has enabled the amelioration of this process leading to the application of this technique in other fields for the betterment of mankind. It not only enables to determine the sequence of the genes in the DNA but also facilitates the identification of any mutations and variations in the gene sequences (Mardis, 2011). Forensic science is the field that involves the investigation of a crime

through the application of various methods and techniques of science and technology. The significance of molecular genetic techniques in forensic cannot be denied as it has been benefitted enormously through molecular genetics. The most recent advancement of molecular genetics which has found application in forensic science is the use of autosomal SNPs as they can provide information about the ancestral genetics of human beings. This has been perceived to serve as a means for the identification of humans and to explore the accomplishment of autosomal SNPs towards the identification of human beings at crime scene.

### **Autosomal Single Nucleotide Polymorphisms**

Single nucleotide polymorphisms are have alleles that are associated with particular physical characteristics in a specific population. Some time if the biological sample is difficult to be analyzed due to in adequate sample (van Oorschot et al, 2010). This difficulty has been overcome by the application of techniques which can amplify the DNA available in reduced quantity or through concentrating on the mitochondrial genome coding region. The most suitable and widely used method for the analysis of the DNA from the degraded sample is the use of Autosomal SNPs. (Budowle and van Daal, 2008).

### **Identification of Victims**

It is not recommended to apply the ancestry based on genetic testing to a diverse population since the traits responsible for the appearance of a person are not restricted to a specific geographical region and are widely distributed throughout the various regions of the world. Therefore, this can be applied or deriving a general idea about the appearance of a person but with particular considerations regarding the applicability of these processes to the cases in which the individuals ancestry belongs to only a single continental region. There is no information to date available regarding the extent of presentation of the traits of the appearance of an individual

that determines the combination of the genes from different continents and regions (Ziętkiewicz et al, 2012).

### **Appearance of a Person**

Considerable amount of knowledge is not present regarding the extent to which the genetic factors determine the appearance of an individual. An insight into the genes which are responsible for the provision of traits to provide information regarding the characteristics such as color of skin, color of hair, traits related to pigmentation such as stature, morphology of hair, and freckles have been recently provided by the genome-wide association studies. The characteristics involved in the incidence of various diseases that are genetically determined such as baldness in male, autosomal dominant woolly hair, and non-syndromic cleft lip can provide information about the genetic fields that are responsible for the variations in appearance of the individuals despite the fact that these traits are not significant for application in forensic investigation. The prediction of a phenotype from a genotype is determined by the extent of genetic effect of SNP on a phenotype and the number of the SNPs that are contributing independently along with the influences that are not related to the genetics. Eye color is the most accurately determined trait from all the traits that have been studied so far (Shimomura et al, 2010).

### **Prediction of Eye Color**

Another limitation with the applicability of DNA for the prediction of various traits that particularly includes the color of eye is dependent on the conceptual understanding of the individuals. For example, there could be assigning of same eye to the categories of different eye color. This could mislead the investigators during the investigation as they could begin searching for the wrong person based on this misleading categorization of the eye color. In order to

overcome this issue a study has investigated the quantitative variations in the color of eye that are based on genetics. This study has observed that there are three eye color genes which are new and the quantitative variation in about 50% of the genes can also be demonstrated through the use data provide by the SNPs (Liu et al, 2009). The development of color charts from these data can be of significant value for avoiding the uncertainties that are caused in the prediction of eye color (Hart et al, 2013).

### **Prediction of Hair Color**

SNPs have also been used for the prediction of hair color with particular emphasis on the prediction of red hair color. This hair color is strongly determined by a single gene which provides significant information regarding red hair color when determined through SNPs. The studies that investigated the application of these types of the predictions in forensics have been performed several years ago. In the investigation of 46 SNPs which were obtained from 13 genes has observed an accuracy of 0.87, 0.93, 0.81, and 0.82 for the prediction of black, red, blond, and brown hairs respectively. It has also enabled to distinguish between the categories of the same hair colors. The dependency of age related to the hair color change might be responsible for the lowest accuracy of prediction of the SNPs for blond hair color (Branicki et al, 2011). The complete variation in the melanin of hair has been observed to be governed by three SNPs. These SNPs were responsible for 76% of these variations. Recently, two of these SNPs have been confirmed to be highly predictive of the hair color. The third gene, among these observed genes, has been recently examined to be related with the biogeographical ancestry instead to be responsible for the prediction of hair color.

## **Prediction of Skin Color**

There is limited availability of the GWA studies for the investigation of the color of human skin. Several genes have been highlighted by these studies but the information related to the determination of skin color based on genes is limited so far. It has been demonstrated by a study performed recently that three SNPs obtained from three genes which are responsible for pigmentation, are accountable for 46% of the variations that are related to color of skin. There is a significant difference in the level of variation caused by the SNPs for the hair color and color of eye with that of the skin color (Valenzuela et al, 2010). This is related with a lowered level of variation of the color of skin in the same individuals. It is not possible to recover the variations of full traits by utilizing the GWA studies for the mapping of genes responsible for imparting skin color to the individuals. This limitation can be determined through GWA studies which utilize the samples from the entire world including various continents to predict the variations in the color of skin based on the differences in the SNPs with regards to different continents. (Sulem et al, 2007). Another study has observed 82% variation in the color of skin by identifying five SNPs responsible for the variation in skin color among the population of the world. It has been considered that these markers would be useful for the prediction of skin colors of various individuals.

## **Prediction of Height and Age**

Height of the human body has also been predicted through the application of SNPs. However, the accuracy of prediction from these SNPs is significantly low which has limited the application of most of the SNPs that are responsible for the prediction of height in forensics. In a recent GWA study that consisted of 180,000 people provided results which identified 180 loci

that are responsible for the height of an individual. These studies have provided several SNPs which can be utilized in future studies for identifying their predictive value (Aulchenko et al, 2009).

Age of an individual can also be predicted through DNA testing. The shrinkage of telomeres and the deletion of mitochondrial DNA related with the process of ageing have found applicability in forensics for the prediction of age of an individual. However, there are certain limitations in their value of practicality. The predictive markers for age related identification of the individuals can be provided through the GWA studies which examine the gene expression related to ageing and patterns of DNA methylation.

### **Research Design**

Secondary qualitative research design has been selected by the researcher for conducting this study. This particular research design has been selected as it is appropriate for enabling the researcher to address the aims and objectives of the study. A systematic review of the literature was performed as it provides an analysis of the studies which have been conducted previously on the topic which is being studied. The researcher has been able to collect the findings of several studies and infer them to develop a comprehensive synopsis of the topic (Barbour, 2010). An analysis of the recent literature has been performed for gathering the information from the recent literature by systematically reviewing the studies (Jolley, 2020). Generation of several themes has occurred as a consequence of analysis of the data provided through review of the literature. This secondary research is based on a systematic review of the studies which have provided an insight in the significance of Autosomal SNPs in forensic sciences through the identification of humans at crime scenes.

A logical search of the relevant terms is performed through the use of Boolean operators. It also results in the exclusion of several items which are irrelevant. Boolean operators are used for eliminating, joining, or expanding the keywords during a search. A combination of diverse key words and the use of capitalize “AND” to restrict the search and “OR” to expand the search when combined is utilized for the search. In order to refine the search, subject heading with map terms were used for the identification of terms from the database thesaurus. Boolean operators were utilized in the search that enabled the researcher to perform the search in accordance with the study topic and narrowing the search. It also saved the time to search the useful research materials and enabled the exclusion of irrelevant items

## **Results**

SNPs have been utilized in cases where the analysis of STRs failed resulted in failure of identification of the humans at the crime scene. In a study which observed six cases of forensics in which the identification failed through the application of STRs utilized SNPs for analysis. The failure of identification was due to the failure of analysis for STRs in the sample. The analysis of the sample was possible through the application of SNPs by using the SNaPshot technique. The samples included in the study were extracted according to the standard protocol. (Köhnemann and Pfeiffer, 2011). Y-SNPs can also be used in forensic investigation for the identification of humans at crime scene. It has been demonstrated that phylogenetic tree of Y-chromosomes can be applied for in forensic which requires to be extensively enhanced. As a consequence of presentation of numerous phylogenetic trees of Y-chromosomes by the researchers, it has become difficult to perform accurate reporting of the results of investigation as it is impossible to relate the results with a phylogenetic tree of Y-chromosomes, which has updated information. Therefore, it has become difficult for the forensic investigators to interpret the results of



investigation based on recent nomenclature. Optimization of the currently available haploid markers of the phylogenetic tree is possible through the data which has been provided by the WGS studies. Only a limited genome set of the Y-chromosome haplogroups is available. Thus, the newly developed tree can be used for the identification of Y-chromosomal trees based on STPs for ensuring their application in forensic investigation (Van Geystelen et al, 2013).

Considering the increased requirement of application of SNPs in forensic sciences for identifying the humans at a crime scene, it is important to develop the techniques and systems which are efficient for providing results that are free from error. A system for the efficient analysis of Y-SNPs has been developed recently. This system was developed by obtaining information from blood sample of six individuals who are not related to each other. Multiplex system of Y-SNPs analysis was designed which ensured effective and efficient analysis of the samples which were artificially degraded. This suggested that such a system can find application in forensic investigation for performing the analysis of SNPs so that it becomes easier to identify a person at the crime scene. Samples that contain mixtures of the DNA can also be analyzed through this system (Zhou et al, 2013). Phenotypic characteristics of the individuals are being predicted through the application of SNPs in forensic investigations. These are valuable markers for the prediction of color of eye, pigmentation of skin, biogeographic origin, and color of hair.

However, the implementation of SNPs for investigation purposes in crime scene human identification is still controversial as there are considerations regarding the legal and ethical implication of SNPs testing in forensic investigation for identifying humans at crime scene.

There are also several limitations that have been observed regarding the application of SNPs for identification of humans at crime scene. These limitations reside in the limited availability of the technologies that can be used for the typing of SNPs and increasing the ability of systems that

are used for typing of SNPs to analyze the biological samples which are highly degraded. Bioethical issues remain a major hindrance in the applicability of SNPs analysis in forensic genetics. Knowledge and ability of the forensic investigators and researchers is essentially required for this purpose to overcome these hurdles in the excessive application of SNPs for identifying humans at crime scene (Gunn et al, 2014). Prediction of eye color of the individuals is also possible through the analysis of SNPs which can serve as a beneficial tool for the identification of criminals and victims at the crime scene. SNPs that were responsible for imparting the color of skin to the individuals were also used. The applicability of these SNPs for the prediction of color of skin and eye were observed to be significant with very low rate of error. This suggested that these SNPs can be applied in forensic analysis for identifying the individuals at crime scene through the prediction of their physical characteristics (Spichenok et al, 2011).

### **Discussion**

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.

The identification of male individuals can also be performed through the utilization of Y-SNPs. A phylogentic tree has been developed for this purpose which has also founded implication in forensic for identifying the criminals and victims through the analysis of the samples which have

been obtained from the crime scene. The Y-SNPs could be useful for the identification of male criminals (Van Geystelen et al, 2013). They can be used for the identification of paternal ancestry which can provide significant information to the forensic investigators regarding the paternal lineage of the criminal or the victim present at the crime scene. This would also be beneficial to reach the criminal by identifying the paternal ancestry of the suspect (Pereira et al, 2010). SNPs are being used extensively for the determination of biogeographical ancestry of the individuals. Recently, it has also found application in the identification of the humans present at crime scene by predicting their phenotypic characteristics and ancestral lineage. Several proficient systems have been designed for performing the analysis of SNPs. This has aided in the identification of criminals and victims at the crime scene.

### **Conclusion**

At present implication of SNPs are being explored in various aspects of forensic sciences for performing the identification of the humans at the crime scene. SNPs have proven to be efficient for predicting the color of eye and hair of the individuals along with the color of skin ( Bulbul O, 2020). This can provide enormous information about the criminal or victim present at the crime scene. This information can be used for identifying the person.

Despite the undeniable significance of SNPs in identification of humans, there are certain limitations which serve as barrier in the implication of SNPs in forensics. These challenges are present in the ethical considerations related with the application of SNPs and the unavailability of the databases with information about them (Gunn et al, 2014). However, these limitations can be overcome and SNPs can be utilized for the identification of humans at crime scene.

## **Recommendations**

Further studies are required to explore the significance of SNPs in the identification of humans at crime scene. These studies should be based on control groups to validate the findings. These studies should focus on the prediction of phenotypic characteristics of the individuals for determining the humans in forensic investigation. However, based on the results of this study, it can be recommended that SNPs should be employed in forensic genetics for identifying the individuals, either victim or criminal, present at the crime scene.

Ethics approval and consent to participate Not applicable.

Consent for publication Not applicable

Abbreviations SNPs:Short Nucleotide Polymorphism ; DNA:Deoxyribonucleic acid ;

STRs:Short tandem ; sjTRECs:signal joint T cell receptor excision circles;GWAS: Genome

Wide Association Study

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**Table 1:** Boolean Logic for the Identification of keywords to Retrieve Relevant Studies

Autosomal	AND	SNPs	AND	Importance
OR		OR		OR
Human		Forensic		Identification
OR		OR		OR
Technique		Crime scene		Genetic