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Forensic DNA Phenotyping: Significance in Criminal Investigations

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Abstract:

In today's present scenario, the forensic investigator faces immense challenges and difficulties to apprehend the perpetrator due to lack of evidence or eyewitnesses. To convict the criminals, the most conclusive evidence is considered to be biological (DNA) evidence. Since criminals have advanced their methods to commit the crime, forensic investigators should also adapt or look for modern techniques to help the court of law to serve justice. One of those modern and most recent techniques is Forensic DNA Phenotyping (FDP). It predicts the external traits of unknown or missing individuals directly from the biological evidence discovered at the scene of crime. FDP can help in providing the lead to the forensic investigator to uncover unidentifiable persons. This application of DNA will be used in forensic casework in a completely different manner from the comparative current DNA profiling which is presently used in the court of law. Presently, no such technique exists which can predict individual-specific appearance accurately. Forensic DNA Phenotyping uses SNPs to determine the phenotype of the individual. It can help in estimating the externally visible characteristics (EVCs) such as hair color, iris color, height, gender, skin tone, and others. Therefore, it develops the biological blueprint of physical traits. This technique can have a great impact in the future and can replace the traditional techniques if provided sufficient funding for research purposes, to develop based on genetically appearance of humans. This will significantly lead to a vast and detailed description of an unidentified person's appearance from DNA, delivering increased value for police investigations in criminal and missing/deceased person cases involving unknowns.

Keywords: *Crime, DNA, Forensic DNA Phenotyping, Evidence, EVCs*

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Introduction

There are abundant genetic variations in the different populations of the world. These variations make up the genome of the individual unique. From the past many decades we are well equipped with the technology and methodology to study these genetic variations. Some of these DNA profiling techniques included are DNA fingerprinting, DNA sequencing techniques, microarray technology, and so on.

As the technology is developing fast in these modern times, there is a need to modify or replace traditional techniques and methods so that it can give more accurate results and is much sensitive than before. One of the advanced technology which has been discovered by scientists is of DNA phenotyping. It can determine the phenotype through the genotype of the individual by using SNP data collected from Genome Wide Association Studies (GWAS) **(Zhang *et al.*, 2014)**. DNA phenotyping technique is more precise and accurate than the previous techniques. It can confirm the gender from the evidence with 100 percent accuracy. It has also 70 percent accuracy in estimating externally visible characteristics (EVCs) like the color of eye, pigmentation in skin and hair, height, and so on.

This technique can have a significant application in forensic science as it will become easy for the investigators to determine the physical features of the culprit without depending upon the known samples. As this technique continues to develop we can determine face shape, the structure of hair, male pattern baldness, an individual's fingerprint patterns, cleft chin, freckles on the skin, skin tone, ethnicity, age, underlying diseases and others **(MacLean and Lamparello, 2014)**.

The FDP (Forensic DNA Phenotyping) can be a useful tool for law enforcement agencies as it is more reliable and more predictive and can help in the exclusion or inclusion of suspects based on the data of this technique which includes the face morphology and external characteristics. Hence, it can be often considered superior to eyewitness testimony, which can be used in the conviction of the wrong person.

Methodology

FDP evaluates single nucleotide polymorphisms (SNPs) in DNA sequences to determine the physical features. Since it is an emerging technique, scientists are still improving it by using many methodologies to make it useful for the society. The most successful methods used to predict physical traits are-

1. Direct Method
2. Indirect Method

1. Indirect methods, genetic variants that are linked with variable expression of the relevant phenotypes are measured and used with the appropriate statistical models to determine its trait value **(Crouch *et al.*, 2018)**.

There are certain traits whose values have been identified. However, these values are based on worldwide genetic testing and have not yet used for forensic purposes. The traits with their respective values are given below **(MacLean and Lamparello, 2014)** -

- Hair Color (Red-10) (Black, Blond, Brown): 11, 12
- Eye (Iris) Color (Blue, Brown): 13-15
- Freckle: 16
- Dimple: 17
- Cleft Lip and/or Palate: 18
- Chronological Age: 19
- Earlobe Attachment: 20
- Adult Height: 21, 22
- Male Pattern Baldness: 23
- Dominant Handedness: 24
- Scalp Hair-whorl Number and Direction: 25

Scientists and researchers have identified various genes which are responsible for a single trait such as **(Marano and Fridman, 2020)** -

- Facial Frontal Contour - OSR1 and WDR35
- Eye Color - HERC2, IRF4, LOC105370627, OCA2, SLC45A2, TYR
- Eye Shape - HOXD1 and MTX2
- Hair Color - EXOC2, HERC2, IRF4, KITLG, MC1R, OCA2, SLC24A4, SLC45A2, TYR
- Nose Shape - SOX9 and DHX35
- Skin Color - ANKRD11, ASIP, BNC2, DEF8, HERC2, IRF4, KITLG, MC1R, OCA2, PIGU, RALY, SLC24A4, SLC45A2, TYR, TYRP1,SLC24A5
- Height - ACAN, DNM3, EFEMP1, FBXW11, GH region, GHSR, GPR126, HHIP, HMGA1, HMGA1, IHH, LCORL, MICA, NOG, NPR3, PML, PPIF, SDR16C5, SOCS
- Facial Features - ADAMTS2, ASPH, C5orf50, COL11A1, COL17A1, CTNND2, DNMT3B, EVC2, FBN1, FGFR1, FGFR2, GDF5, LRP6, PAX3, POLR1D, PRDM16, RAI1, RELN, ROR2, SATB2, SEMA3E, SLC35D1, TP63, UFD1L, WNT3
- Craniofacial Formations - WDR35 and SOX9
- Baldness - AR/EDA2R, EBF1, HDAC9, TARDBP, 20 p11

2. In indirect methods, ancestry markers are used to match with the phenotypic trait of the person by using statistical models to identify its trait value.

Usually, the direct method is preferred over the indirect method because many geographical populations have a mixed race and ethnic background. Therefore, it becomes difficult to identify the true ancestors of the person **(Koops and Schellekens, 2008)**.

Versatility of Parabon's Snapshot

One of the US based companies named Parabon NanoLabs Inc. has designed the software Snapshot under the funding of the US Department of Defense. It uses advanced MLA (Machine Learning Algorithm) and in-depth data mining to develop Snapshot.

The Snapshot is currently used to identify the following-

1) DNA Phenotyping

2) Forensic Art Enhancement

3) Facial/Skull Reconstruction

4) Ancestry Determination

5) Kinship Inference

6) Genetic Genealogy

1) DNA Phenotyping

The Snapshot DNA Phenotyping System uses SNP data from a person's sample to predict ancestry and physical traits. The prediction is made with a measure of confidence, including those that can be excluded with high confidence.

FDP System accurately predicts genetic ancestry, EVCs, and face shape in individuals from any ethnic background. It generates data on an Illumina® microarray scanner and with a 200ng quantity of DNA, it can give 100 percent accuracy. This software helps in estimating the biogeographical ancestry of the individual. By using 2.5ng of DNA sample, it can give an accuracy of 98 percent while using 1ng of DNA sample it gives an accuracy of 95 percent. Such quantity is generally encountered in forensic cases **(Kayser and Manfred, 2015)**. Though, they are still working to make their software sensitive to detect ancestry and physical traits in forensic samples which are degraded in both quality and quantity.

2) Forensic Art Enhancement

Snapshot forensic art services provide a means to add information such as age, body mass index (BMI), the presence of facial hair, eyeglasses, piercings, etc into a Snapshot composite which is decoded from non-coding DNA sequences. The forensic artists who are trained in using this software can do the digital facial reconstruction by using the structure of bones to enhance a Snapshot composite.

3) Facial/Skull Reconstruction

The Snapshot Facial Reconstruction Service provides the most completely formed recreations of ante-mortem appearance from the skeletal remains. Forensic artists use markers to apply the depth of the tissues manually on the deceased's skull which are bases on DNA ancestry and approximate weight of the body to predict the shape of the face from cranial morphology. A final composite is then formed by digitally combination of the two predictions.

4) Ancestry Determination

The Snapshot uses high-dimensional SNP data and uses two different approaches for ancestry inference-

I. Principal Component Analysis

II. Statistical Clustering

I. Principal component analysis (PCA) combines correlated variables into a smaller set of uncorrelated variables that describes the variance present in the original data. Each point will represent a single individual in the reference database, with a location on the plot determined solely by their DNA, after which the points are marked with the color, according to the subject's known ancestry **(Crouch *et al.*, 2018)**.

Individuals with a mixed background like African-American will show ancestry intermediate between the parent groups (African/European). In this way, it becomes possible to locate an unidentifiable person to a vast population group by identifying his or her genotypes.

II. Statistical clustering uses a reference database to define a set of populations, against which an unknown person's DNA is compared to determine its proportional participation in each. This allows for a mixture, even between populations that have not previously been observed.

In the first stage of analysis, subjects from around the world are included. In the second stage, only subjects from the inferred continent(s) are included.

5) Kinship Inference

Snapshot provides results about the relationships between individuals or distantly related individuals with accuracy by using SNPs to detect relatedness out to 9th-degree relationships (fourth cousins). This gives forensic investigators valuable knowledge about the DNA samples found at the scene of crime. The method has proven to be highly accurate while maintaining a very low false-positive rate **(Greytak, 5, 6)**.

6) Genetic Genealogy

Genetic Genealogy is a combination of genetic analysis with genealogical research to study family history. It also uses the Snapshot Kinship Inference software to study if the individuals are related or unrelated. It can be paired with Snapshot DNA Phenotyping to further reduce the list of possible matches. This can aid forensic investigators to apprehend the criminal.

Limitations

There are certain limitations which are faced by scientists due to which FDP cannot be accepted by the legislatures of various countries. One of the limitations of this technique is that it cannot generate data with complete accuracy if there is a mixed racial profile of the individual. The person is not restricted to the particular geographical location and will show various other characteristics including ethnicity and race. Another challenge is that if the country accepts this technique in the court of law then there will be no privacy of the individual **(Toom *et al.*, 2016)**. The government can access the personal information of the person like ancestral background, the medical information of the diseases, etc. Databases of such information can threaten one's life if it is misused for personal purposes. This is one of the main reasons that many countries have not accepted this technique **(Koops and Schellekens, 2008)**.

Another limitation of the FDP technique is that there is a change in environmental factors that affect the functioning of the genes. These effects can be correlated with the changes in the physical features and therefore, it will be difficult for scientists to estimate a trait as a mutation or naturally occurring in the population.

Further, there are many overlapping markers present in the genome which causes hindrance in understanding and identification of markers particularly present for the phenotype. Moreover, the EVCs which can be given a trait value are limited **(MacLean and Lamparello, 2014)**.

The forensic samples are always contaminated and degraded, to determine the trait values from the SNP markers from those samples is a challenge. In addition, the improper handling, collection, and packaging of forensic samples makes it further difficult to analyse them. If scientists overcome these limitations then FDP will be the most successful technique in the field of forensic of recent times **(MacLean and Lamparello, 2014)**.

Feasible Future Advancements

Technology has the great potential to become more advanced and give better results than before. It requires more progress in other domains rather in the facial morphology, ancestral background, and externally visible phenotypic features. Such improvement in the technique will make it more accurate and precise.

With the help of some experiments, scientists have found that FDP can locate and predict relevant DNA markers via Next Generation Sequencing (NGS)

technology. Recent technologies like NGS and DNA Phenotyping have the ability to change the forensic analysis. Some geneticists have also used the current advanced methods such as cytosine methylation of DNA to determine the age of the individual **(MacLean and Lamparello, 2014)**.

One of the main advantages will be in the field of forensic science that it can help in repairing the damaged DNA samples by a complete understanding of one's genome.

Conclusion

We can conclude that this new emerging technology of FDP can prove to be a great asset to the country. If many softwares like snapshots are designed then the crime rate can be reduced exponentially. Software like Gene Identification via Phenotype Sequencing (GIPS) is under testing for the same purpose and many other softwares are at the developing stage.

But the main challenge lies in the acceptance of this technique in law systems of various countries. So far, Europe and Australia have some laws on DNA phenotyping as they are the most advanced countries in DNA forensics. In fact, Australia has closed a case with the help of FDP. Moreover, Netherland is the only country that is unambiguously allowing the use of DNA for phenotyping.

Other countries like Israel, South Africa, Brazil, etc have less infrastructure for the development of DNA forensics and are using traditional techniques to some extent. Belgium, Germany, and the 3 states of the US namely, Indiana, Rhode Island, and Wyoming, have completely prohibited the use of DNA for deriving physical traits as they find it ethically wrong to extract the personal data of an individual without their consent. They are using DNA to identify only the gender of the person.

Due to these moral and regulatory issues, the global acceptance of Forensic DNA Phenotyping seems to be a reality in a distant future.

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