

Forensic Phenotyping



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Abstract

Forensic DNA phenotyping [FDP] is a new approach that uses DNA to create probabilistic inferences about a person's observable traits. It is an amazing fact that helps us to learn about something from the object that they left behind at the scene of crime. The most discarded items like cigarette buds, chewing gums, Tobacco residue and bidi. From this discarded waste, it helps to extract a trace amount of DNA evidence to create a DNA profile of a person which helps to reconstruct a person's face. This process can be done, with the identification of hair colour, height, eye colour, sex, age, and even morphology of the face, and the ancestry of criminal and accused persons with the help of their genetic constituents. A person's genotype can help to develop a particular phenotype. By taking this point into consideration forensic DNA phenotyping can analyze or predict a person's possible face or physical appearance. By implementing this technology into the forensic science for the low enforcement agencies and police department to reduce their work. And for directing the line of investigation and getting little bit idea about the suspect 108 nanograms of DNA are present in the few microliter of saliva which was present on the chewing gum. And of which about 0.5 nanograms of DNA are required for standard forensic analysis. This will be acts as the witness against criminals, and provide an investigative leads to trace unknown persons who are unidentifiable with current comparative DNA Profiling this intelligence application of DNA marks a substantially different forensic use of genetic materials rather than that of current DNA profiling presented in the courtroom.

Keywords: Ancestry; DNA Phenotyping; DNA; Forensic Genetics; Evidence; Nanograms; Microliter

Abbreviations: FDP: Forensic DNA Phenotyping; EVCs: External Visible Characteristics; Aims: Ancestry Informative Markers; BMI: Body Mass Index

Introduction

One of the most effective methods for both criminal and personal identification is forensic phenotyping. DNA samples are frequently discovered at crime scenes due to criminal or suspect error, which allows us to quickly profile the offender and identify them using a newly developed technique known as the Forensic DNA phenotyping method. Genotyping or DNA sequencing data may be used for DNA phenotyping, which is the technique of predicting an organism's phenotype only from genetic data. A person's DNA may reveal visible qualities like hair colour, eye colour, facial shape, skin tone, sex, and many more by looking at the sequencing of their genomes.

In 2013 Artist Heather Dewcy-Hagborg created a mask of strangers based on the genetic material left behind at the scene of crime typically DNA found at the crime scene are very less in

amount, but it is enough foot the identification of suspect. In one microliter of saliva contain a 108 nanogram of DNA, of which 0.5 nanogram of DNA sample are required for the standard forensic analysis. 150 cells are required to yield 1 nanogram of DNA and about 37 trillion cells in the human body. Fingernails of the human consist of 40 nanograms of DNA and about 5 nanograms of DNA are present in the single hair of human beings. and on an average about 50 hairs are shaded in a single day, so this is the advantage for the low enforcement agencies and forensic scientist to make a carrier in the field of forensic DNA phenotyping.

Forensic DNA phenotyping is prediction of human appearance from a trace samples like blood, semen, saliva and other bodily fluid, this method also provides way's to the investigation for low enforcement agencies without a known suspect but the most difficult problem face by the scientist it docents stand

in the courtroom it is the biggest challenges in front of scientist. this method is also providing an investigative lead to trace unknown person who are unidentifiable with current comparative DNA profiling this intelligence application of DNA marks a substantially different forensic uses of genetic material rather than that of current DNA profiling presented in the courtroom. The molar body, which is the visible and palpable body that is readily disclosed to the expert's gaze, was the centre of attention in the 19th century.

An abundance the focus on the body has, however, been enhanced recently by molecular- level biometric technologies and graphical tools that make the inside of the biological body readable while also enabling its anatomization, breakdown, modification, and amplification at the molecular level (Rose, 2001, 2007). The biological samples gathered at crime sites are processed to create a DNA profile that will be compared to the profile of the suspect, aiding the investigation to pinpoint the perpetrator and the crime scene, or even ruling out suspects [1].

Main Body:

Hair Colour:

Hair colour contributes significantly to our overall visual appearance and our personalities. Thus, hair colour products offer us the option of having the hair colour of our choice nowadays. The fundamental factor in our body to determine our hair colour is determined by the amount of a pigment called melanin in the hair. "Eumelanin and pheomelanin are the two pigments that play a significant role in how hair colour depends. [MC1R] melanocortin 1 Receptor. Melanins are pigments derived from an amino acid called Tyrosin. (MC 1 R) is the gene responsible for hair colour determination in human beings. The main difference observed in hair colour is the result of two types of melanin: brown / black eumelanin and red / yellow pheomelanin [2]. This variability in hair colour has probably arisen in Europe, evolving from a dark ancestral colour, resulting from human mating preferences [3].

Most studies do not contemplate the sampling of younger individuals, or question adult subjects about distinct phenotypes in early childhood. Therefore, predication models are only elaborated with phenotypic information observed in adults, without taking into account information observed in adults, without taking into account informative markers for age- dependent phenotypes, partially explaining the lower accuracy value for blond hair. There is a study done with young individuals found that hair darkening usually occurs between 6 and 13years of age and that Hlrisplex model incorrectly predicts hair phenotypes for those individuals who were blond only during early childhood, advising for the need to identify new markers that could reduce this error rate [4].

One more comparison study was done in hair examination methodologies in which study was conducted to investigate the

accuracy between two methods of hair analysis: PCR-STR DNA analysis and microscopic comparison analysis. Standard sets of public hair were collected from participated volunteers, and unknown sets were generated from the taken samples. Three out of five (60%) of the hairs analyzed produced full DNA profiles that were correctly matched to the standard sets. DNA analysis was inconclusive (partial or no DNA profile) for two out of five (40%) of the samples. In contrast, the microscopic comparison analysis correctly matched four out of five (80%) of the samples to the standard sets but mis-identified one out of five (20%) of the samples. These results reinforce the practice of preliminary microscope hair examination in narrowing down a set of hairs for DNA analysis. Microscopic comparison analysis is sufficiently reliable to remain a rapid and inexpensive method for forensic hair analysis [5].

Skin Colour:

Skin tone is an adaptive feature that has been well researched in humans; it is one of the most variable pigmentation phenotypes ever observed. The variation in skin pigmentation is thought to have developed as an evolutionary reaction to the strength of UV light in various planetary locations. Increasing selection pressure would maintain dark skin present in places closer to the equator with greater luminous intensity (high UV), whereas lesser selective pressure would be present in regions farther away from the equator with lower luminous intensity [6]. In the past several years, significant progress has been made in identifying the genes responsible for the variance in skin tone in a variety of geographical and temporal populations. This evolutionary factor makes the genotype/phenotype associations in mapping studies in mapping studies difficult, as well as resulting in correlations that only apply to a specific population group. While association found in admixed populations did not have the same discriminatory power in more homogeneous populations, such as Europeans [7]. In a crucial period, several tests conducted on a homogenous population were unable to distinguish between the skin tones of disparate groups of Asian, African, and Native Americans [8]. Additionally, a variety of cases of skin colour adaptability in both worldwide contemporary humans and ancient homo erectus are included, along with an explanation of the causes, timing, and processes of skin colour adaptation in various populations. Taking into account this evolutionary obstacle, a global prediction model was developed based on 36 markers distributed among 16 pigmentation genes [9].

Eye Colour:

Eye colour has been thought to exhibit the greatest variations among human traits, ranging from royal blue to dark brown or black colours, through intermediary tones like grey, hazel, yellow, and green. A recent study shows the Iris plex model for eye colour prediction was analyzed in 238 voluntaries of Italian ancestry to evaluate their possible applicability as a tool of DNA intelligence in forensic investigation, the result of the

study confirms the previous findings from several different populations showing once again that the iris plex system predicts blue and brown eye colour with high accuracy while it is inefficient in the prediction of intermediate eye colour [10]. However, since the accuracy in their forecasts is still significantly lower when compared with blue and brown eyes, intermediate eye colours are still a concern, mandating more study to establish novel genetic variations [11]. An aspect contemplates gender as a possible influencing factor in the determination of eye pigmentation. It has been observed that women tend to have darker eyes than men in some of European countries [12-13].

The Iris Plex technique is acceptable for predicting the blue and brown eye colours in the tiny Iraqi population, according to Nihad A.M. Al-study Rashid's on eye colour prediction. In the Iris plex system, the prediction of intermediate eye colour is dependent on two SNPs that, according to the study, represent nearly all samples with wild type and have disappeared due to effective change in both SNPs. Six SNPs were genotyped in the study, and it is thought that this would have had an impact on the phenotypic variation in eye colour. Therefore, if additional SNPs are genotyped in the larger Iraqi population, prediction accuracy can be increased [14]. More research should be performed to evaluate this link because no genetic element has yet been identified to account for this variation.

Height:

Only a significant minority of genes have been identified as being relevant to human height as of 2008. Continuing association studies, which were carried out in 2008 and 2010, the number of genetic markers increased to 180, and then reached over 700 markers in 2014 [15,16]. Even the number of height-related variations has significantly increased. There are still no significant values for prediction tests. A recent study found accuracy values of 65%, while the most recent research was unable to increase this value to >75%, highlighting how complicated this feature may be and how many SNPs need to be uncovered [17].

Since there are multiple gene variations that affect height (a pattern of inheritance known as polygenic inheritance), it is challenging to estimate a child's final height. Children ordinarily grow to be nearly tall as their parents, but various combinations of alleles might lead siblings to be of diverse heights. This is because one inherits these variants from one's parents. Other natural processes that may also be controlled by heredity, such as hormones, have an impact on height, albeit their exact functions are not thoroughly known. Height is affected by environmental variables, such as a mother's diet during pregnancy, whether she smoked, and her exposure to hazardous chemicals, in addition to genetic and biological determinants.

Although studies on immigrant families have shown that moving to a country with better access to nourishing food, healthcare, and employment opportunities can have a significant impact on the height of the next generation, it is possible that some

ethnic differences in height are not genetic in nature. In addition, there are variables other than genetics that may contribute to human height, including gestational (placental characteristics and maternal health factors including diet, diseases, and medicines), hormonal, and environmental (nutrition and lifestyle) factors, particularly throughout infancy.

Age Estimation:

Age estimate is a core part of forensic phenotyping being one of the external visible characteristics (EVCs) that can be employed to learn more about a sample source. Age-related DNA methylation may be exploited as an accurate age estimate owing to the discovery of epigenetics. The age approximation of an individual has been established to benefit from epigenetic research employing DNA methylation detection methods, which take a different approach from those previously depicted here (SNP typing mostly). In the curriculum of forensic science investigations, the assumption of the donor's chronological age plays a crucial role.

It gives investigators leads in the adventure for an unrecognized accused person in a variety of situations: it is crucial for human identification from skeletal remains; the age of a person in question determines whether the constitution applies; and it can be crucial in immigration cases where the identity and age of a person are uncertain. Previous age categorization systems included morphological examinations of bone characteristics [18]. DNA methylation changes throughout an individual's life-levels of methylation increase in childhood and then decrease after reaching adulthood [19].

Sequence analysis of bisulfite-converted DNA now seem to be the most promising method for calculating chronological age, according to methylation-based forensic investigations, even though it still requires more DNA than is often present in forensic samples. While a typical forensic sample only comprises around 10 ng of DNA, methylation studies conducted outside of the forensic sector sometimes employ 200ng or more of input DNA. To make this technique more widely used in everyday forensic work, further research will need to concentrate on reducing the DNA input necessary for methylation analysis. Consistency of protocols and methodologies will also be necessary [20].

Sequence analysis of bisulfite-converted DNA is now the most widely used method for identifying CpG sites. This method involves treating single-stranded genomic DNA with sodium bisulfite, which delaminates unmethylated cytosine to uracil while leaving methylated cytosine unaltered. Additionally, to concentrated DNA hyper methylation of CpG islands, genetic sequence DNA hypo methylation has also been seen as adults age [21].

Facial Features:

When analysing phenotyping, one of the main aims is to anticipate the face shape of all EVCs in order to get a glimpse of the final «DNA facial composite.» The distances between facial

landmarks, such as the width of the lips, the height between the eyes and the face, and the cephalic index, are used to study the morphology of such a face. *FACES* as a software application that gives you a lot of possibilities for recreating someone's face characteristics. Some of the genetic markers associated with facial features are initially found in syndromes and facial deformities diseases studies (such as cleft palate, cleft lip, and other craniofacial dysplasia's). Some of these markers are then correlated to craniofacial development and consequently linked to the normal variation of facial shape [22]. The strategies used by Claes were based primarily on data from admixed populations; individuals use a first step in which sample ancestry and gender are used to create a base-face; data from 24 SNPs will then be used to add information about the nose, lips, face roundness, jaw, chin, and supraorbital crest to this primary face. Significant correlations with face thickness, brow width, eye distance, columella inclination, nose bridge width, nostril width, and mouth shape were also identified in another research [23].

Ancestry:

An individual's ancestral origin can be revealed by certain DNA markers, revealing precise information on their biogeographic contributions (Africa, Europe, Asia, and Amerindian). Ancestry informative markers (AIMs) can therefore be used to infer a person's ancestry, giving information to support possible witnesses, or even revealing new details about crime scene evidence [24]. An individual's genealogy cannot, however, be the only feature taken into consideration when judging how they look. One must Acknowledge the disparity between ancestry and the erroneous person of race: a person's percentages of ancestry will not always correspond to how they look.

Application and Real Cases:

The Edmonton Police Service sent a press release on April 18, 2019, summarizing the facts above and asking for information regarding the suspect, someone that they described as being 5'4» and wearing a black toque, jeans, and jumper or hoodie. He was said to speak with an accent. He was thought to have left the area heading west. After a prolonged investigation that uncovered no witnesses, CCTV, public tips, or DNA matches, investigators decided to work with Parabon Nano Labs, a Virginia-based DNA technology business that specializes in high-tech DNA analysis services. DNA phenotyping, which predicts physical characteristics and ancestry based on unidentified DNA evidence, was the service utilized in this instance. In criminal investigations, police organizations utilize the company's Snapshot ® DNA Phenotyping Service to reduce suspect lists and produce leads. For the connected person of interest, Parabon made trait predictions using DNA evidence from this inquiry (POI). Each subject's ancestry, eye colour, hair colour, skin tone, freckling, and facial shape were individually predicted. A «Point - in - time» composite was created by integrating these physical characteristics to show what the POI would have looked like at age 25 and with an average body mass index (BMI) of 22.

Because age and BMI cannot be established from DNA, these default values were utilized [25]. (Police use DNA phenotyping in unsolved sexual assault). Using the company's Snapshot ® DNA Phenotyping Service, law enforcement officials may create suspect lists and generate No match was established in DNA databases after a woman in Florida (USA) was sexually attacked in broad daylight in 2010. Seven years later, the police hired a private DNA phenotyping business, which produced a face composite that identified the suspect as a guy with light brown complexion, brownish-hazel eyes, and black hair. A suspect with the same traits was discovered by police at the crime scenes nearby property, a nature refuge, according to the new DNA face composite.

Hugo Giron-Polanco was detained following a voluntary sample donation because a DNA comparison revealed a 1:400,000,000,000 match probability between his STR profile and that of the semen sample taken from the victim [26]. A decision to allow police in the German state of Bavaria to evaluate DNA samples and estimation ancestry as well as hair, eye, and skin colour was developed on May 15, 2018. The Netherlands, France, the United Kingdom, Canada, and a total amount of US states have all employed DNA predictions, despite the fact that some of these nations lack clear regulations on the practice [27]. Computers may eventually be able to examine DNA-based composites produced from biological evidence against government databases or even images from social media. This approach will be able to narrow down the topics of interest, even if it does not identify a perfect match, which will be extremely beneficial in the future.

Recent Trends in DNA Phenotyping:

One of the most intriguing areas of current genetic research, which has applications in many other disciplines, is DNA phenotyping. It might, however, lead to more applications that raise ethical concerns. It is clear that as this technology has advanced in the future, stringent standards and practices will need to be put in place on a global level to avoid possible abuse of this innovative and exciting framework by governments and law enforcement government agencies. DNA phenotyping is primarily used in the field of forensic science. It is used to narrow the pool of potential suspects or identify remains by learning about the ancestry and appearance of a person. DNA phenotyping has frequently assisted policemen in the past year and a half in locating suspects or human remains.

One of the most well-known incidents was the armed robberies of Candra Alston and Malaysia Boykin in Columbia, South Carolina. Kenneth Canzater Jr. was accused of the crime in 2017 after police in 2015 used a composite picture that was the first to be created exclusively from a DNA sample. A mug shot was created by Parabon Nano Labs and was made from a digital mesh representing the expected face morphology, which was then layered with textures representing the expected colour of the eyes, hair, and skin. Archaeology is another discipline where

DNA phenotyping has applicability. The method may be used by researchers in this subject to learn more about the appearance of our ancestors.

Forensic Science Current Developments:

With all the genetic phenotyping materials are described here, it can be concluded that creating a collection of genetic markers for discovery and analysis that can reliably predict the largest percentage of human EVCs is now closer than ever, and that a full «DNA face composite» is currently within reach. In addition to finding new links between physical traits, more research must be done to verify the data collected from many worldwide populations and to make sure that none of the associations observed are caused by ancestry or another population background. However, the excellent statistical correctness of most of this research makes them suitable for practical application in forensic measures, despite the numerous ethical and legal problems that still pervade this subject.

Apart from monozygotic twins, DNA analysis for forensic investigations is predicated on the notion that every person is genetically unique. By directly comparing the genetic profile of short tandem repeats acquired from biological samples of unknown origin to a reference sample profile, DNA retrieved from biological samples can individually identify this material. The prerequisite for a reference sample for comparison is one of the main drawbacks of this method. Studies looking at the connection between certain polymorphisms and specific phenotypic traits are multiplying, and the results are encouraging for forensic sciences. Forensic DNA phenotyping is the method of determining externally visible characteristics (EVCs) from biological materials, such as height, facial features, iris, and hair colour, and male baldness pattern (FDP). Therefore, without the necessity for a reference sample for comparison analysis, FDP provides significant information about the subject to which a specific biological sample belongs. It is important to consider certain moral and legal issues so that this new technology does not incentivize racial or ethnic discrimination.

Conclusions:

A properly defined legal framework should govern the use of the DNA phenotype in forensic investigations. To guarantee the privacy of any personal information that a genetic profile may supply and to prevent discrimination and physical simplifications, it should also be governed by establishing control of data management and interpretation, not only by forensic scientists but also by legal professionals. With all the genetic phenotyping materials are described here, it can be concluded that creating a collection of genetic markers for discovery and analysis that can reliably predict the largest percentage of human EVCs is now closer than ever, and that a full «DNA face composite» is currently within reach. In addition to finding new links between physical traits, more research must be done to verify the data collected from many worldwide populations and to make sure

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