H70 Forensic DNA Phenotyping (FDP): A Prediction of Human Externally Visible Traits in Missing Person Identification

Matteo Fabbri, MS, University of Ferrara, Ferrara 44121, ITALY; Letizia Alfieri, MD, University of Ferrara, Ferrara 44121, ITALY; Omar Bonato, MD, University of Ferrara, Ferrara 44121, ITALY; Mauro Coppone, MD, University of Ferrara, Ferrara 44121, ITALY; Paolo Frisoni, MD, University of Ferrara, Ferrara 44121, ITALY; Rosa Maria Gaudio, University of Ferrara, Ferrara 44121, ITALY; Elena Lucenti, MD, University of Ferrara, Ferrara 44121, ITALY; Chiara Marini, MD, University of Ferrara, Department of Legal Medicine, Ferrara 44121, ITALY; Matteo Marti, PhD, University of Ferrara, Ferrara 44121, ITALY; Margherita Neri, MD, PhD, University of Ferrara, Ferrara 44100, ITALY*

Learning Overview: The goal of this presentation is to inform attendees of a new "DNA intelligence" tool that is approaching forensic biology, due to its ability to predict Externally Visible Characteristics (EVCs) from biological material, such as that found at crime scenes, in disaster victim identification, or in cases of missing person identification. EVC prediction from forensic samples, or from body parts, is expected to help concentrate police investigations toward finding unknown individuals at times when conventional DNA profiling fails to provide informative leads.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by attempting to prove and underline the utility of FDP in missing person identification cases. Due to the lack of data in literature, this work involved the application of the HIrisPlex system for eye, hair, and skin color prediction to 20 Italian missing person identification cases previously studied. In order to evaluate the accuracy of the multiplex tested, 20 cases for which victim pictures or portraits were available were chosen.

Predicting EVCs using informative DNA molecular markers has started to become a rapidly developing area in FDP. The most relevant forensic cases for DNA-based EVCs prediction would be those in which the evidence DNA is useful for describing a person's physical appearance from skeletal remains. This study evaluates the HIrisPlex DNA test multiplex for the simultaneous prediction of eye, hair, and skin color in 20 Italian skeletal remains from 20 different missing person identification cases in which identification was previously performed using the conventional Short Tandem Repeat (STR) -based method. In all 20 cases, victim pictures or portraits were utilized as a reference in order to test the reliability and accuracy of the DNA-based EVCs prediction.

The DNA-based system used consists of a SnaPshot® multiplex assay targeting a total of 41 Single Nucleotide Polymorphisms (SNPs) involved in skin, eye, and hair color prediction[1]. A total of 20 different bone samples (14 femurs and 6 tibias) were evaluated and processed. Bone samples were cleaned chemically, using diluted bleach, and irradiated with Ultraviolet (UV) light for 30 minutes prior to grinding into a fine powder. Genomic DNA was obtained from 0.5g of bone powder using the QIAamp® DNA Investigator KiP. The DNA extracts were quantified using the Investigator QuantiPlex™ HYres Kit. The reactions were carried out in a Rotor-Gene® 5-plex System according to the manufacturer's instructions. Polymerase Chain Reaction (PCR) amplification of all 17 SNPs was performed in a single multiplex PCR assay, in a final volume of 10µl, as described by Chaitanya et al.[1] The amplified PCR products were purified with Thermo Scientific® ExoI and incubated at 37°C for 45min and 80°C for 15min. Single Base Extension (SBE) was carried out for all PCR products simultaneously in a single multiplex reaction using 2µl of the purified PCR products. Finally, the purified SBE products were analyzed on the ABI® 310 HID Genetic Analyzer. Gene Mapper® ID-X v1.0 software program was used for the allele calling and analysis of the results. Prediction of eye, hair, and skin color was performed using a web interface at https://hirisplex.erasmusmc.nl/ allowing the retrieval of individual prediction probabilities for three eye color, four hair color, and now five skin color categories from HIrisPlex genotype input data of 41 SNPs. Each victim picture's iris, hair, and skin color were subjectively and objectively determined for all 20 skeletal remains analyzed. For the subjective determination, examiners classified victims' pictures in the same color category (i.e., iris, hair, and skin color) for 84% of the documents examined.

Study results showed overall prediction accuracies of 91.6%, 90.4%, and 91.2%, respectively, for iris, hair, and skin color at the 0.7 threshold. The sample set had only two inconclusive results as compared to the IrisPlex database. This is likely due to the fact that these samples showed an intermediate eye and hair color. In conclusion, these results demonstrate the robustness and reliability of the HIrisPlex genotyping system. Utilization should be encouraged for the prediction of eye, hair, and skin traits from DNA in missing person cases and in outside anthropological applications to determine eye, hair, and skin color of deceased persons via analyzing skeletal remains or body parts.

Reference(s):

1. Chaitanya L., Breslin K., Zuñiga S., Wirken L., Pośpiech E., Kukla-Bartoszek M., Sijen T., et al. The HIrisPlex-S system for eye, hair and skin colour prediction from DNA: Introduction and forensic developmental validation. *For. Sci. Int.: Genet.* 2018, 35, 123-135.

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*Presenting Author