FORENSIC DNA PHENOTYPING THROUGH MASSIVE PARALLEL SEQUENCING

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Massive parallel sequencing, MPS, (also known as next generation sequencing) has changed the face of genomic research and is expected to change the way forensic analyses are performed in the future. The ability of this technique to produce results from hundreds to thousands of variants in a single run is an ideal tool for forensic researchers to expand the assays available to assist law enforcement investigations, particularly in the area of physical appearance prediction.

Forensic DNA phenotyping is a type of 'biological eye witness' as it has the ability to predict externally visible characteristics from DNA left at a crime scene. Recent research in studying the genes and, more specifically, the variants that are responsible for physical appearance characteristics have made incredible strides in the last few years, in particular the advancement of eye, hair, and skin color prediction. Work on eye and hair color prediction using 'HIrisPlex' has allowed accurate predictions of blue or brown eye color with a precision greater than 95%, and of hair color with a precision of approximately 75% for blond, brown, black and red categories [1]. Skin color prediction is soon to follow suit providing a full categorical pigmentation profile.

However, current tools and prediction models are limited by technology and the number of variants needed to be genotyped in the biological assay. Therefore, right now, only categorical color prediction systems have been optimized and these can be subject to interpretation (i.e. one's idea of 'light brown hair' may not be uniform). In order to move towards continuous or quantitative pigment prediction, the MPS technology opens many new doors to individualized pigment prediction, as the number of DNA variants required to predict continuous color is vastly larger: hundreds to thousands. The greater the variant capacity – the higher the prediction accuracy, provided the fundamental knowledge of the additional pigmentation genes is known. In an effort to move towards more individualized continuous pigmentation prediction profiles, the ability to combine all known pigmentation markers in an easy to use single multiplex assay, as done here using MPS technology, is highly beneficial to the forensic community. It is also one step closer to combining all known physical appearance prediction markers, beyond pigmentation, into one library preparation for massive parallel sequencing and the ultimate biological sketch.

1. Walsh, S., et al., *Developmental validation of the HIrisPlex system: DNA-based eye and hair colour prediction for forensic and anthropological usage.* Forensic Sci Int Genet, 2014. 9: p. 150-61.