A189 Validation of SNPs to Predict Pigment- Related Features in Diverse Populations

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After attending this presentation, attendees will understand some principles of pigmentation, including some components of its pathway. Attendees will be aware of how single nucleotide polymorphisms (SNPs) may affect eye, hair, and skin color.

This presentation will impact the forensic science community by describing pigment-related features based solely on DNA analysis.

An essential component in the identification of human remains is documenting the decedent's visible characteristics, such as eye, hair, and skin color. However, if a decedent is decomposed, or only skeletal remains are found, critical, usually externally visible information is lost. It is presently not possible to use genetic information from recovered, fragmented DNA samples to reveal such visible characteristics due to the lack of markers that are significantly correlated with visible traits.

Human genomes demonstrate 99.9% identity, while the remaining 0.1% accounts for variations between individuals, which include single nucleotide polymorphisms (SNPs). Based on sequencing results, it has been estimated that the human genome contains at least 11 million SNPs, of which most are silent and do not contribute to a phenotype. However, some have functional consequences. Since there are so many SNPs, it is challenging to find those which are directly responsible for human traits.

Melanin is the main pigment of eye, hair, and skin color and its synthesis depends on multiple genes and factors, such as age, drugs, diseases, and environmental conditions. Pigmentation is therefore considered a complex trait. Furthermore, only minimal correlation exists among eye, hair, and skin color, within the European population, where blue and brown-eyed individuals can have all shades of neutral hair colors. In other geographical regions; however, populations with darker skin tones tend to have darker eye and hair colors. To reduce complexity, previous studies focused on a single trait in one population, such as the eye color in Europeans, or on few genes, including MC1R, SLC45A2, OCA2, HERC2, ASIP, and SLC24A5, whose products are associated with melanin synthesis or its localization, to identify SNPs that correlate significantly with eye, hair, and/or skin color.

Presented at this meeting is the validation of several SNPs for their predictive value to describe eye, hair, and/or skin color of individuals. A predictor, comprised of several SNPs that were found to correlate well with these traits is created and applied on over 600 samples. Importantly, these samples were donated from non-related individuals of various populations, including European descendants, African-American individuals, Asian dark (Indians), Asian white (Japanese and Chinese descendants), and mixed populations and are thus well representative among the various populations. The authors will present the outcome of this validation for the pigmentation traits by focusing on accuracy and call-rate as well as applicability to the diverse populations.

The ability to utilize DNA analysis to predict visible pigment-related features of unidentified human remains has the potential to assist in the identification of missing persons and unidentified human remains by increasing the amount of information available which can be entered into the National Missing and Unidentified Persons System (NamUs) and the National Crime Index System.

Pigmentation, SNP, Identification