A17 Using SNPs to Predict Hair Pigmentation in Individuals of European Ancestry

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After attending this presentation, attendees will become more familiar with genetic methods for predicting hair pigmentation. The genetics of human hair pigmentation will be briefly reviewed, as well as its forensic utility. In addition, the effectiveness of SNP analysis in predicting hair pigmentation in individuals of European ancestry will be discussed.

This presentation will impact the forensic science community by discussing an assay for inferring the lightness or darkness of an individual's hair color. Data from such an assay could be useful in legal investigations.

An assay that could help provide a physical description of a person from a DNA sample would be helpful when eyewitnesses are either not available or have conflicting reports. In addition, such an assay could help identify missing persons who cannot be identified through traditional means. On average, medical examiner and coroners' offices handle roughly 4,400 unidentified missing persons each year, with approximately 1,000 remaining unidentified after a year.[1] In these instances, a lack of reference DNA can hamper a successful identification. To address this, researchers have investigated genetic loci that produce phenotypic differences among individuals. This could aid in identifications by producing a physical profile of an individual. Thus far, assays have been developed to predict an individual's eye color,[2] ancestry,[3], and red hair color[4] using single nucleotide polymorphisms (SNPs) in conjunction with population allele frequencies. Since the existing hair pigmentation assay can only differentiate red and non-red, it would be useful to have an assay to predict other hair colors.

Hair pigmentation, along with skin and eye pigmentation, is determined by melanin. There are two types of melanin; eumelanin is the black/brown pigment, while pheomelanin is the yellow/red pigment. Melanin is synthesized in melanocytes, and the number, size, and distribution of melanocytes contribute to the shade differences observed in hair, skin, and eyes.[5] Strong correlations exist between hair and/or eye color and the SNPs examined. Therefore, the genotypes of each SNP are expected to correlate with hair pigmentation.

Ten SNPs located in nine pigmentation genes were chosen for analysis, including *SLC24A4*, *KITLG*, *OCA2*, *TYR*, *IRF4*, *MATP* (*SLC45A2*), *HERC2*, *TYRP1*, and *SLC24A5*. DNA samples were collected, along with data on background characteristics (including hair pigmentation) and ancestry informative markers. Two primer extension multiplexes were developed and optimized for genotyping the samples. The genotypes were tested for linkage to hair pigmentation by admixture mapping, and a model for predicting hair pigmentation was designed.

The proposed model was then tested by analyzing an additional set of samples blind. The results were compared to the individuals' original reported hair color and pigmentation measurements, and the accuracy of the chosen SNPs in predicting hair pigmentation was determined. The effectiveness of the ten SNPs individually, along with interactions between pigmentation and ancestry, were determined

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