B173 Forensic Use of Hair Pigmentation Gene SNPs to Predict an Individual's Hair Color

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After attending this presentation, attendees will become familiar with genetic methods that may be useful for predicting hair pigmentation. Attendees will receive a basic overview of the genetics of hair pigmentation, and how this knowledge is useful in forensics. The effectiveness of SNP analysis in predicting human hair pigmentation will be discussed.

This presentation will impact the forensic community by increasing the understanding of pigmentation genes, and detailing a forensic assay for inferring an individual's hair coloration. Such data could be quite useful in a legal investigation.

In the absence of video or witness identification, DNA-predicted hair pigmentation may be used to help create a physical profile of an offender and aid in police investigations. Additionally, it could be useful in developing a physical profile of remains that cannot be identified through conventional means. As of May 2007, DNA profiles generated from crime scene evidence reached a total of 177,870 in the National DNA Index System Forensic Index,[1] and CODIS had over 49,400 hits.[2] According to The 2004 Bureau of Justice Statistics' Census of Medical Examiner and Coroners' Offices, approximately 4,400 unidentified human decedents are discovered every year, about 1,000 of which remain unidentified.[3] In both instances a lack of reference DNA poses a limitation to successful identification. To circumvent this, DNA analyses that do not require reference samples may be extremely useful. The analysis of genetic loci that produce obvious phenotypic differences among individuals has the potential to directly aid in identification. Among these, single nucleotide polymorphisms, or SNPs, in conjunction with population allele frequencies, can allow for inferences of physical characteristics of an individual. These can be summarized to produce a physical profile or "fuzzy photo" of an unidentified individual. Furthermore, SNP analysis can be carried out using short amplicons to aid in the genotyping of degraded DNA, and can be conducted using high throughput techniques, making it well suited for automation and databasing.[4]

This presentation will discuss the forensic use of SNPs to predict hair color based on several genes that contribute to pigmentation. The pigment melanin determines the natural color of skin, irises and hair in humans. Melanin is synthesized and packaged into granules called melanosomes. The degree of pigmentation depends on the type of melanin and the size, shape and density of melanosomes.[5] Many genes influence the melanin synthesis pathway, and thus human pigmentation. There has been recent research into the genetics of hair pigmentation, but little on the predictive value of a genotype in deducing an individual's hair color. Strong correlations between skin, eye, and/or hair color and the SNPs examined are known to exist; therefore genotypes of each SNP are expected to correlate with hair pigmentation.

Eight SNPs located in three different pigmentation genes (*SLC24A5, SLC45A2* and *MATP*) were selected for study. DNA samples were collected, along with data on background characteristics (including hair pigmentation measurements) and ancestry informative markers. A multiplex SNP primer extension assay was developed and optimized to carry out the genotyping, and the samples were analyzed for the SNPs of interest. Using an admixture mapping approach, the genotypes were tested for linkage to hair pigmentation. A model for predicting hair pigmentation from the eight SNPs studied was designed.

The proposed model was then used to predict hair pigmentation for an additional set of samples, which were assayed blind. The results were compared to the individuals' original hair color descriptions and pigmentation measurements and the accuracy of the eight SNPs in predicting hair pigmentation was determined. The effectiveness of the SNPs individually and collectively in predicting hair pigmentation will be discussed.

References:

1 U.S. Department of Justice. "NDIS Statistics". *CODIS Homepage*. 2007. <http://www.fbi.gov/hq/lab/codis/clickmap.htm>.

2 U.S. Department of Justice. "Measuring Success". *CODIS Homepage*. 2007. <http://www.fbi.gov/hq/lab/codis/success.htm>.

3 Ritter, Nancy. "Missing Persons and Unidentified Remains: The Nation's Silent Mass Disaster". U.S. Department of Justice, Office of Justice Programs, National Institute of Justice. The NIJ Journal. 2007 Jan;256:2-7.

4 Sobrino B, Brión M, Carracedo A. "SNPs in forensic genetics: a review on SNP typing methodologies". Forensic Sci Int. 2005 Nov 25;154(2-3):181-94.

5 Sturm, RA, et al. "Human pigmentation genetics: the difference is only skin deep". Bioessays. 1998 Sep;20(9):712-21.

Hair Pigmentation, SNP, DNA

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