Program: The Prediction of Human Physical Appearance from DNA
Speaker: Susan Walsh, Ph.D., Assistant Professor, IUPUI
Introduced by: Malcolm Mallette
Attendance: 149
Guests: Greg Brophy, Amy Manders, Hunter Heath, Mark Jones, David & Mag Zauner, Merrell Porteous, Eve Perlitern, Gordon McLaughlin, Charles Russell
Scribe: Benny Ko
Editor: Bonnie Carter

Forensic DNA Phenotyping: The Quest for a Biological Portrait

DNA phenotyping is basically designed to generate a DNA-based portrait of a person and use it to solve a forensic and/or anthropological void. Such issues could be the crime scene bio-evidence that does not match to known DNA profiles, to assess and corroborate with an eye-witness account, to aid in a police search of an individual with little personal data or image, and to lend a face to an unidentified deceased or even a previously unmet ancestor.

The science first requires the collection of a large cohort of cases and controls (n>1000), then a genome-wide association study (GWAS) based on single-nucleotide polymorphism (SNP) arrays is conducted. The association signals are detected, mapped and replicated and finally, biologically validated.

Following that, different models for prediction are developed and tested for validity. Currently, the most understood traits are that of the eye, hair and skin color and are considered to be three categorical prediction models. For example, using the IrisPlex system that studies 6 SNPs, a

95% prediction accuracy of blue and brown eye color can be expected. For hair color prediction, things get a little more complicated. Here, 11 single SNPs and 2 combined sets of variants leading to a total of 22 SNPs are found to be involved. It allows the prediction of 4 categories of hair colors, blond, brown, black and red with accuracy values ranging from 0.81-0.93. Interestingly, blond is harder to predict accurately in an adult as hair color often changes as a child grows.

Skin color prediction is by far the most complex of the three organ-systems studied. In one study, 14185 SNPS in 281 genes were reported involved in skin color expression. For practicality, only a subset of these SNPS and genes are studied in skin color prediction.

The HIrisPlex-S (HPS) system is an all-in-one pigmentation prediction that incorporates the evaluation of eye colors, hair colors, and skin colors. Eye color prediction is correct in 80% of cases. The accuracy increases to 96% when only two colors are considered, blue and brown, and the intermediate colors are not included. Hair color prediction is 77% accurate on average. Skin color prediction accuracy is 80% on average.

Other appearance traits that could be predicted from DNA studies and GWAS are lifetime aging, head hair structure, i.e., hair texture, and hair loss pattern. Future promises include the prediction of height, body weight, and facial morphology, but as yet, the genetic knowledge is limited.


Dr. Susan Walsh