DNA PHENOTYPING: PREDICTING PIGMENTATION TRAITS IN A UNITED STATES POPULATION

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DNA phenotyping is the ability to determine physical external characteristics based on genotype analysis. This work will prove valuable for forensic casework where DNA from biological evidence is limited in providing probative information for the investigation. Physical characteristics of the donor such as hair, skin, or eye color, may present investigational leads when the conventional STR profile of the unknown DNA sample does not match any suspects or victims nor yield any hits in a DNA database. It may also be useful for the identification of missing persons or victims of mass disasters to provide investigators with the most likely appearance of the unknown individual.

Pigmentation traits analyzed here include iris and skin color, both complex genetic traits determined by several genes and resulting in highly polymorphic phenotypes. The IrisPlex system was developed in the Netherlands and is a SNP genotyping assay combined with a statistical model for predicting eye color. The system genotypes at six eye color informative single nucleotide polymorphism (SNP) loci and using the genotype data, a multinomial logistic regression model was developed and used for predicting the probability of eye color into three categories: brown, blue, and intermediate. Previous work focused on evaluating the accuracy of the IrisPlex system's prediction component in a North American (U.S.A.) population, and the evaluation of a new prediction model approach, Bayesian network analysis. Furthermore, a subset of the U.S.A. sample population was genotyped at three additional SNPs that may prove informative for predicting between skin color phenotypes.

Digital photos of each individual's right eye were collected and objective color classification was done by quantifying the iris color based on RGB color components. The iris color was evaluated on a scale of a single numerical value derived from the three values, called the iris melanin index, and the skin color was evaluated from an extract of the same photo based on the average RGB color value and classified as light, medium, or dark.

The iris color prediction results based on the U.S. sample population using the same prediction parameters do not have the same level of accuracy or prediction power as the original study, the maximum prediction accuracies of the IrisPlex system after population allele frequency adjustment was 58% and 95% brown and blue eye color predictions, respectively, and 11% for intermediate eye colors. For skin color prediction, three SNPs found in literature to be informative for predicting light and dark skin color were tested. At least two of the three additional SNPs were found to be informative whereas the variant at each loci indicative of darker skin color phenotype was found in samples classified as medium or dark skin color. However, not all samples classified as dark skin color were found to have the informative variant, which can lead to false negative predictions. The three SNPs were evaluated for prediction accuracy in a subset of the U.S. sample population collected.