A QUANTITATIVE APPROACH TO MEASURING EYE PIGMENTATION FOR GENOME WIDE ASSOCIATION STUDIES

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DNA Phenotyping is an up and coming area within the forensic DNA analyses community that has many possible applications, from casework to anthropological studies. However, while there are currently several tools that aid in the prediction of categorical phenotypes, they ultimately require further refinement to improve the accuracy of the profile they deliver. Increasing our fundamental knowledge of new genes that contribute to a phenotype, even minor contributions, has the ability to build more accurate prediction systems. One way to find these 'predictive' genetic markers is to perform genome-wide association studies using novel phenotyping approaches on several hundreds of individuals.

For this study we concentrated on eye color. As previous studies have already found many of the major genes that contribute to blue or brown-eyed individuals (predominately *HERC2*), elucidating genes that have smaller effects is more difficult. Essentially, as our knowledge of these minor contributing genes increases, it will be possible to begin moving past categorizing eye color as simple colors such as blue and brown and instead to start classifying in terms of real quantitative pigment, such as the amount of pheomelanin and/or eumelanin.

Therefore, in order to better identify novel genes that help determine iris color, we have developed and implemented a computer-based automated approach that captures not only quantitative color information from iris imagery, but also location-based color information in an effort to elucidate the genes responsible for pigment deposition. This approach allows us to potentially identify genes involved in a wider variety of eye colors that better reflect individual iris variation. Ideally, we hope to translate our findings using the refined iris phenotyping pipeline to improve eye color prediction models on a quantitative scale.