A167 Validation of IrisPlex in the United States: A DNA-Based Tool for the Prediction of Eye Color

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After attending this presentation, attendees will understand the concept of DNA phenotyping, especially relating to iris color, the application of the IrisPlex system and its prediction model component, alternative approaches for quantifying iris color, and other possible prediction methods.

This presentation will impact the forensic science community by providing information regarding 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 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. S DNA phenotyping is the ability to determine physical external characteristics solely based on genotype analysis. Iris color is a complex genetic trait determined by several different genes resulting in highly polymorphic phenotypes. The IrisPlex system was developed in the Netherlands and is a multiplex SNP genotyping assay combined with a statistical model for predicting eye color. The system targets six eye color-informative, Single Nucleotide Polymorphisms (SNP) and using the genotype data, a multinomial logistic regression model was developed based on minor allele frequencies and is used for predicting the probability of eye color into three categories: brown, blue, and intermediate.

This work focuses on the developmental validation of the IrisPlex system and to evaluate the accuracy of the system's prediction component on a North American population. Validation of this work was done amplifying the same SNPs using the SNaPshot chemistry single base extension method. Capillary electrophoresis was performed on the ABI 3500 genetic analyzer.

The results of the IrisPlex study indicated predictions with greater than 90% accuracy of blue and brown eye color based on a European (Dutch) population. The prediction results based on the North American sample population so far, using the same parameters in the IrisPlex model, do not have the same level of accuracy or prediction power, especially with the blue color. All but one sample tested from the North American population so far is heterozygous at the rs12913832 SNP locus, which has been found to have the highest association with blue and brown eye color predictions when in the homozygous state. The highest probability predicted with the North American samples collected so far is 56% for blue eye color. These results are reasonably expected as North America is a highly admixed population compared to the European populations previously studied, which is where most eye color variation originates.

The parameters of the statistical regression model need to be adjusted for the differences in the allelic and genotypic frequencies between the two populations, and this is one of the goals of this work as a way of improving the prediction accuracy. In addition to validating the method using a North American population, other methods for prediction and quantitation of iris color, possibly to achieve more accuracy with the intermediate color category, are considered and evaluated. One such method for eye color classification is quantification based on separation of the iris color into red, green, and blue (RGB) and luminosity values to create a numerical color scale. Association of that value with the genotype data may be used for prediction purposes.

Iris Color, SNPs, IrisPlex

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