A166 Implementing SNPs Into Forensic Casework: An Assay and Interpretation Models to Predict Ancestry and Eye Color

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After attending this presentation, attendees will be informed of a new Single Nucleotide Polymorphism (SNP) assay that can be used to predict ancestry and eye color of an unknown individual, also covering developmental validation, steps of individual lab implementation, and interpretation of results.

This presentation will impact the forensic science community by understanding that when an STR DNA profile obtained from crime scene evidence is not consistent with known or database profiles, no further information is available from genetic evidence. If forensic laboratories were to implement this assay, then further analysis targeted at inferring the ancestral origin and phenotypic characteristics of the perpetrator could provide valuable investigative information. Results of this analysis would aid in prioritizing suspect processing, corroborating witness testimony, determining the relevance of evidence to a crime, and ultimately increase the ability to identify individuals related to the crime scene.

The existence of population-specific SNP variation found throughout the world can be used to predict the ancestry of an unknown individual, and, in some cases, can also predict externally visible characteristics (EVC). A panel of 50 SNPs showing strong association to pigmentation phenotypes and/or ancestry has recently been determined and an assay with which to type these SNPs using the single base extension (SBE) technology has been developed. Evaluation of this panel found it to be sensitive and robust, similar to current STR typing methods. All the primer sequences, protocols, and analysis parameters (including GeneMapper ID and GeneMarker panels/bins) are publicly available. This panel can be implemented with minimal investment, as it uses the same equipment and similar methods already in use in forensic DNA casework laboratories.

Also to aid in implementation, an ancestry interpretation method has been made available, using a U.S. sample set and the open source web-based application Snipper. By adding the genotypes from an unknown sample and running Snipper with the known sample set, users will receive likelihood ratio results, which have been shown to reliably predict the ancestry of an unknown individual. Further, if the individual is determined to be of European ancestry, results of this evaluation show that a freely available excel-based prediction model can be used to indicate eye color. The 50 SNPs selected were also based on their ability to provide information on hair color and skin melanin content; however, at this time the number of individuals with known phenotype and corresponding genotype data for these SNPs is insufficient to generate reliable prediction models. Funding to increase the size of this U.S. sample database is being sought.

Lastly, evaluations are ongoing to determine if other forensic DNA results (STR, YSTR, mtDNA) can be integrated to increase the ancestry prediction ability. Using the free, excel-based Omnipop, a method of evaluating the random match probabilities of an individual's STR profile in different populations that could be used to predict ancestry has been developed. Also under development is the incorporation of the YSTR haplotype worldwide distribution using a web-based haplogroup predictor. These methods have been tested on a U.S. sample set to determine if combining results from other markers strengthens or weakens the SNP ancestry prediction. Overall, such a model could be incorporated into forensic casework to maximize the information obtained from an unknown sample.

SNP, Ancestry, Phenotype

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