DEVELOPMENT OF A SNP ASSAY PANEL FOR ANCESTRAL ORIGIN INFERENCE AND INDIVIDUAL SOMATIC TRAITS

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When an STR DNA profile obtained from crime scene evidence is not consistent with known or database profiles, 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.

Single Nucleotide Polymorphisms (SNPs) are the most common form of genetic polymorphisms present in the human genome. A large number of SNPs have alleles that are associated with particular populations and/or correlated to specific somatic traits. For example, the Duffy (*FY*) blood group phenotype FY (A-B-) is lacking the receptor for *P. vivax* malaria, reducing susceptibility to malarial infection in Sub-Saharan African populations. This adaptation to malarial infection only occurs in Sub-Saharan African populations and is useful as an Ancestry Informative SNP (AISNP). An example of Phenotype Informative SNP (PISNP) is found in the melanocortin-1 receptor gene (*MC1R*) which is involved in the transfer of melanin, affecting hair and skin pigmentation. Mutations reducing the efficiency of this receptor (among other pigmentation genes) represent an adaptation to the reduced amount of UV light available to humans as they migrated away from equatorial regions.

Single Base primer Extension (SBE) is an efficient and sensitive technology that can type over 30 SNP loci throughout the genome in a single reaction. We have developed a panel of robust and sensitive SBE assays which include 100 SNPs associated with ancestry and somatic traits, and are evaluating candidate SNP results for predictive power on volunteers of known ancestry and phenotype. The goal of this project is to provide an analytical tool using technology currently available in forensic DNA laboratories, which could be implemented in the form of a kit and used on casework as needed.