A165 Estimating Genetic Ancestry Using SNP Analysis

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After attending this presentation, attendees will understand how utilizing SNP technology in a specific assay can be applied to crime scene evidence to obtain genetic ancestral information.

This presentation will impact the forensic science community by discovering new forensic DNA applications available for criminal investigations in helping to reduce the number of potential suspects in an unsolved or cold case.

The utilization of many worldwide DNA databases, such as CODIS, can be an essential tool in modern criminal investigations. Unfortunately, when an evidentiary DNA profile does not provide a viable suspect subsequent to a database search, the investigator may be left with little forensic direction. The use of ancestral testing can be a potential option to obtain additional information regarding the donor of DNA left at a crime scene. However, there are limitations to ancestry testing due to the potential that a person's overall ancestry may be wrongly assumed with the use of haplogroups. The mtDNA or YSTR testing methods are only evaluating a small, selected portion of a person's genomic ancestry. To assist in these critical situations, Sorenson Forensics developed Investigative LEAD™; a Single Nucleotide Polymorphism (SNP) based DNA test designed to estimate genetic ancestry against a model of five genetically distinct, putative parental populations. The populations and the reference samples representing them are as follows: Western European (HapMap CEU, Northwest European descent residing in Utah), West Sub-Saharan African (HapMap YRI, Yoruba from Ibadan, Nigeria), East Asian (HapMap CHB from Beijing, China), Indigenous American (compilation of samples identified as being from populations indigenous to North, Central, and South America including Maya, Pima, Karitiana, Surui, and Arawak descent), and the India Subcontinent (HapMap GIH, Gujarati Indian descent residing in Houston, TX). In addition, the Investigative LEAD assay covers all autosomal chromosomes and the test markers are associated with ancestral informativeness so that an accurate representation of the human genetic anthropology is obtained. Our method uses 190 SNP Ancestry Informative Markers (AIMs) chosen from their scored ability to specifically differentiate between the five reference populations using Principal Component Analysis (PCA) as the comparative analysis tool and includes some markers identified as informative in previous genetic ancestry estimation publications. The method uses fluorescence-based polymerase chain reaction reagents to provide qualitative detection of targets using post-PCR endpoint analysis. As a modified approach to standard genotyping, this system miniaturizes the reactions down to 33 nanoliters for cost efficiency and high throughput. The data analysis uses a Principal Component Analysis (PCA) and a proprietary algorithm based on the program FRAPPE. The method calculates affinity levels of an individual DNA sample and then compares that to at least a hundred randomly selected subsets of individuals from the reference populations. Background interference is calculated simultaneously and is used to estimate confidence intervals based on a calibration that was effected using thousands of worldwide individuals. The effects of inhibitors, species specificity, sensitivity, non-probative casework samples, and a comparison of extraction methods from the developmental validation will be presented to demonstrate that the test is robust and viable for the forensic sample types frequently encountered in criminal investigations. The test is capable of providing valuable information regarding the genetic ancestry of the donor of crime scene DNA evidence, which can subsequently aid in reducing a pool of suspects in the investigation. Another application for this technology will be to assist in the determination of the possible contributor of skeletal remains.

SNP, Analysis, DNA

