A112 Utilizing Ultra-High-Density SNP Arrays to Analyze Forensic Mixtures: An Operational Assessment

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After attending this presentation, attendees will learn about direct solutions for multiple source forensic DNA samples.

This presentation will impact the forensic science community by providing a precise solution to forensic DNA mixture samples.

The use of Ultra High Density SNP Arrays (UHDSAs) for forensic investigations has transitioned from proof of concept to an accomplished tool for fighting crime. Much like Moore's law for computing, during the last two years, the UHDSAs have increased in size from several hundred **contributors** is mostly wrong. For example consider a mixture with thousand SNPs to 2.5 million, while the cost per SNP has decreased. In

fact the power of this tool is approaching the capability to analyze all the known variations in the human genome. Technically, the UHDSA provides a precise measure of the genetic variation which allows for a more direct assessment of differences between individuals and populations than is possible using current sieving technology (capillary electrophoresis).

The use of UHDSA was validated according to the ISO17025 guidelines. As part of those validation studies population databases for Caucasians, African-Americans, Hispanics, and Asians were generated for the greater than 1 million SNP loci in the Illumina HumanOmni1- Quad system. Like STRs there are allele frequency differences between populations. But because the loci are bi-allelic those differences can be pronounced. For example, in the Caucasian population the B allele frequency for locus rs1002005 is 0.032 while in the African-American population the frequency is 0.985. While more extreme, it none the less is reflective of the population differences found in the frequency databases of STRs. Second, the mixture interpretation algorithm used to assess membership in a forensic mixture is sensitive to the frequencies of the genetic variants in the reference population. The data show that this sensitivity is especially important when less than 1,000 SNPs are used but negligible when greater than 500,000 SNPs are used.

The real test of the power of SNPs is their operational use in deciphering forensic mixtures. Comparison of STR results using sieving technology and SNP results utilizing ultra-high-density SNP arrays demonstrates that the UHDSAs provide a much more precise manner of mixture interpretation. Included in that precision is a statistical assessment of identity as compared to a CPE interpretation.

A review of mixtures from forensic samples using both STRs and UHD SNP analysis demonstrates that the power of 1.4 million SNPs provides a decisive and precise method of mixture analysis that is impossible with STRs.

Algorithms, data, and forensic interpretation methods are proprietary.

SNPs, Forensic Mixtures, DNA Analysis