

UTILIZING ULTRA-HIGH-DENSITY SNP ARRAYS TO ANALYZE FORENSIC MIXTURES: AN OPERATIONAL ASSESSMENT

Kevin C. McElfresh, Kristin Stanford, and Ron Sosnowski
Casework Genetics, 13580 Groupe Dr, Ste. 301, Woodbridge, VA 22192

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 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 capillary sieving technology.

The staff at Casework Genetics has validated the use of UHDSA according to the ISO17025 guidelines. The data from those validation studies is beyond the scope of this presentation. However 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 STRs frequency databases. 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.

Algorithms, data, and forensic interpretation methods are proprietary.