IDENTIFICATION OF AN INDIVIDUAL IN COMPLEX FORENSIC MIXTURES USING SNP MICROARRAYS

Sheri Ayers MS[1], Lev Voskoboinik[2], Ariel Darvasi PhD[2], Aaron LeFebvre PhD[1]
[1]Cellmark Forensics
[2]Hebrew University of Jerusalem

The identification of an individual in a complex DNA mixture poses a challenge to forensic DNA analysis. Statistical approaches have been developed for such purposes using short tandem repeat (STR) kits, but the small number of loci in these kits limits the utility of these kits for interpretation of mixtures with greater than three individuals. In this study, we have demonstrated proof-of-principle of our unique approach for interpretation of complex mixtures using standard “off-the-shelf” high density single nucleotide polymorphism (SNP) microarrays. By selecting a panel of 3000 SNPs with relatively low minor allele frequency (MAF, <0.15) for analysis, our approach is sufficient to determine the presence or absence of an individual in a complex mixture of several contributors with high statistical significance, even when the contribution of that individual is low or when the mixture contains up to 10 different individuals. Using this approach, when 5ng of total DNA was subjected to whole genome amplification (WGA), individuals contributing as little as 5% to a mixture were detected using this approach. Similarly, individuals who contributed as low as 2% to a mixture were detected when 25ng DNA was used. Finally, even in the presence of common inhibitors found in typical forensic-type samples (*i.e.* hematin, humic acid) as well as when DNA is highly degraded, an individual can be identified in a complex mixture with high statistical significance using this approach. In conclusion, our study demonstrates that in principle our statistical approach works with real forensic-type samples using currently available standard high-density SNP to identify individuals in complex mixtures. Future work with a focused low-density forensic SNP microarray is proposed that may be more applicable to real forensic type samples with limiting amounts of genomic DNA.