B163 Evaluation of SNPs as Tools in Human Identity Testing

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After attending this presentation, attendees will learn the utility of single nucleotide polymorphisms (SNPs) as human identity markers. The attendee will learn about different classes of SNP markers, typing technologies, and how SNPs compare with STR markers.

This presentation will impact the forensic community and/or humanity by reporting on the utility of SNP markers to the DNA testing community. SNP markers can provide valuable complementary roles in human identity testing. The typing of coding region mtSNPs and small autosomal panels of SNPs for typing degraded DNA are two examples of where SNPs can benefit the forensic community.

SNPs have the potential to play a helpful role in human identification testing. The small PCR amplicon sizes associated with SNP typing technologies make SNPs attractive for typing degraded DNA or other low copy number situations. SNP can be useful in combination with STRs for resolving complex paternity issues (e.g., incest), identifying victims of mass disasters where insufficient family references are available and possibly inferring population of origin. SNPs located in the coding region of the mitochondrial genome have been used to separate common HV1/HV2 mitotypes thereby extending the power of mtDNA testing (1,2). SNPs located on the Y chromosome have been evaluated for ethnicity prediction and individual sample discrimination (3).

Various SNP typing platforms exist, but at this time there is not a universally agreed upon platform for SNPs and human identity testing. Currently researchers are typing SNPs with multiplex allele specific primer extension (ASPE) reactions. The assay is comprised of an initial step of PCR followed by primer extension and subsequent fragment separation and detection by capillary electrophoresis. ASPE multiplex panels can routinely type 6-12 SNPs in a single tube and have reported to go as high as 35 SNP markers.

Important considerations for SNP markers are the larger number required to equal the discriminatory power compared to traditional STRs, their inability to resolve complex mixtures, issues related to databasing new loci, and the availability of a standard analysis platform. However, in appropriate situations SNPs can be useful as a supplementary tool complementary to STR markers.

Methods and Materials: A total of 70 bi-allelic (C/T) SNP markers have been typed for 189 U.S. samples. Amplifications were performed in 6-plex panels. Amplicons between 59 –108 base pairs were generated. The SNP markers were typed using multiplex ASPE assays and capillary electrophoresis. An 11-plex ASPE assay for typing coding region SNPs that helps resolve the most common Caucasian mitotype was developed and run on samples previously screened by Roche linear arrays for a common HV1/HV2 mitotype. Multiplex assays consisting of 3 ASPE multiplexes and 5 commercial hybridization multiplexes were used for typing 50 Y-SNPs. The 50 Y-SNPs were typed for 229 U.S. African American and Caucasian samples.

Summary of Results: Novel multiplex SNP assays have been developed for typing various classes of SNP markers on U.S. population samples. Subset panels of the 70 autosomal SNP markers have been used to successfully type DNA from shed human hairs. Results for mtSNPs and Y-SNPs allow for an evaluation of the practical utility of various SNP markers in a human identity context. Samples typed by commercial and novel multiplex STR panels allow for a direct comparison of SNP and STR markers.

Conclusions: Practical and inherent characteristics of SNP markers will prevent them from replacing traditional STR typing methods. However, SNP markers can provide valuable complementary roles in human identity testing. The typing of coding region mtSNPs and small autosomal panels of SNPs for typing degraded DNA are two excellent examples of where SNPs can benefit the forensic community.

References:

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