B25 The Development of a Highly Informative, Hierarchical Multiplex SNP Typing System to Predict Ethnogeographic Ancestry Using Pyrosequencing Technology

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After attending this presentation, attendees will be informed of new highly discriminating Y-SNPs and their potential use, in combination with mtSNPs and autosomal SNPS, for predicting the ethnogeographic ancestry of an individual.

This presentation will impact the forensic community and/or humanity by showing attendees the potential uses of Y-SNPs, whether alone or in combination with STRs, as a tool in human identification.

The ability to determine ethno-geographic origin of an individual can potentially provide descriptive information of an unknown individual who deposited a biological stain at a crime scene, therefore serving as a genetic eyewitness. This can potentially be accomplished with the careful selection of population-of-origin specific Y chromosomal single nucleotide polymorphisms (Y-SNPs), mitochondrial (mtSNPs), or autosomal SNPs. Y chromosomal SNPs (Y-SNPs) are increasingly becoming important due to their paternal inheritance, lack of recombination, abundance, and low mutation rate and have been investigated for use in determining population structure. Potential forensic applications of Y-SNPs include their use in predicting the ethnogeographic origin of the donor of a crime scene sample, inclusion, or exclusion of suspects of sexual assaults (the evidence of which often comprises male/female mixtures and may involve multiple perpetrators), paternity testing, and identification of non- and half-siblings. Currently, many of the well characterized Y SNP markers do not differentiate between the large distributions of individuals belonging to the more common haplogroups such as the major European, African, and Asian derived haplogroups R1b3, E3a, and O. Additionally, more diverse populations, such as the Hispanic/Latin groups cannot be differentiated with the only current set of well defined Y-SNPs. The use of several recently phylogenetically defined Y-SNP markers that have the ability to differentiate between sub-populations within common Y-SNP haplogroups, and the use of mtSNPs and autosomal SNPs that can assist in distinguishing between many European, African, Asian, and Hispanic/Latin individuals within particular admixed haplogroups are reported here.

During the course of this study M222, a sub-R1b3 marker rarely used, and found several individuals that possess this polymorphism and also possess Y-STR haplotypes identical or derived from the 17 marker Irish Modal Haplotype (IMH) described by Moore et al were evaluated. Since IMH and IMH-1 individuals possess almost identical STR haplotypes, it is necessary to choose more informative STRs for individual identification of individuals within this haplogroup. How it is possible to replace a battery of Y-STRs with a certain Y-SNP, such as M222, allowing only the most discriminating STRs to be analyzed for individual identification of individuals within a particular haplogroup will be shown.

Several SNP genotyping methods are available, but many do not allow high-throughput multiplexing or are not sensitive. Pyrosequencing is a sensitive and reliable method that can be useful when a hierarchical multiplexing strategy is used. Pyrosequencing can consistently detect picogram quantities of DNA with a nested PCR amplification approach. The development of a sensitive, hierarchical multiplex system for use with pyrosequencing technology, incorporating the most informative Y-SNPs and enhancing their potential to discriminate between sub-populations by including mtSNPs and an ancestry informative autosomal SNP will be described.

Y-SNPs, Y Chromosomal Haplogroups, Pyrosequencing

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