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THE USE OF RECENTLY PHYLOGENETICALLY DEFINED Y-SNPS IN A TYPING SYSTEM TO PREDICT ETHNOGEOGRAPHIC ANCESTRY USING PYROSEQUENCING TECHNOLOGY

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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. Y chromosomal single nucleotide polymorphisms (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. Unique mutations within the non-recombining region (NRY) of the Y-chromosome (mainly SNPs) have created population specific paternal lineages (commonly called haplogroups) that have persisted throughout human history. It is unclear the extent to which SNPs will augment STRs as the primary method of identification in forensic science but potential forensic applications of Y-SNPs include their use in predicting the ethnogeographic origin of the donor of a crime scene sample, as well as for 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 the identification of non- and half-siblings. Currently, many of the well characterized Y SNP markers do not differentiate between the large numbers of individuals belonging to the more common haplogroups such as the major European and African derived haplogroups, R1b3 and E3a. Here we report the characterization of several previously phylogenetically undefined Y-SNP markers that have the ability to differentiate between sub-populations within these Y-SNP haplogroups, therefore increasing the discrimination potential for Y-SNPs.

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. We have shown that pyrosequencing can consistently detect picogram quantities of DNA with a nested PCR amplification approach. We will describe the development of a sensitive, hierarchical Y-SNP typing system for use with Pyrosequencing technology, with the incorporation of the most informative population-relevant Y-SNPs.