EXAMINATION OF Y STR LOCI TO ENHANCE PATERNAL LINEAGE FORENSIC ANALYSES

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The Y chromosome short tandem repeat (Y STR) markers have become a mainstay of forensic biological evidence analyses. The paternal inheritance, smaller effective population size, and lack of independence between Y STR loci can reduce diversity and may yield greater population substructure effects on a locus-by-locus basis compared with the autosomal loci. Studies are needed on forensically-relevant populations to determine empirically what population substructure effects exist and what methods might be applicable to correct for those effects when estimating the rarity of a Y STR haplotype. The United States population is composed of a wide array of population subgroups that harbor different degrees of genetic variation. Population studies are necessary to assess the genetic variation of forensically-relevant markers so that proper inferences can be made about the rarity of DNA profiles.

This study examined 16 Y STRs in four sample populations found within the United States: Native Americans (from Alaska), Caucasians, African Americans, and southwestern Hispanics. The following Y STRs were typed: DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS456, DYS458, DYS635, DYS448, and Y GATA H4. In addition, these samples were typed for the 15 STR loci (which includes the 13 core CODIS loci).

Forensic population genetic and statistical issues addressed were: 1) the degree of diversity at the locus and haplotype level; 2) determination of the loci that contribute more so to haplotype diversity; 3) using these four sample populations, the effects of population substructure on forensic statistical calculations of the rarity of a Y STR profile; and 4) whether there is support for the proposition of multiplying the frequencies of an autosomal STR and Y STR profiles to provide a composite estimate of the rarity of the combined profiles.

DNA from buccal swabs taken from the four sample populations was extracted using the EZ1 Advanced XL instrument by Qiagen. The purified DNA was quantified using the Quantifiler™ Human DNA Quantification Kit DNA (Applied Biosystems, Foster City, CA). Y STRs were amplified using the reagents contained within the AmpFℓSTR® Yfiler® PCR Amplification Kit (Applied Biosystems). Autosomal STRs were amplified using reagents contained within the PowerPlex® 16 HS system (Promega Corporation). DNA fragments were separated by capillary electrophoresis in an Applied Biosystems 3130xl Genetic Analyzer (ABI). Genotype data were analyzed using GeneMapper® ID v3.2 software.

All four population samples were highly polymorphic at the haplotype level for the 16 Y STR markers. Even though Native Americans demonstrate reduced genetic diversity compared with other major US populations, the genetic variation within Native Americans is still quite high based on forensically-relevant genetic markers. Population parameters, statistical applications, and interesting null alleles and duplications will be presented.