

EXAMINATION OF NOVEL 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 their gene 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 empirically determine the power of discrimination, the effects of population substructure, and the methods that might be applicable to apply when estimating the rarity of a Y STR haplotype. Previous studies using the AmpF&STR® Yfiler® PCR Amplification Kit (Applied Biosystems, Foster City) have shown that major population groups are highly polymorphic for 17 Y STR loci haplotypes. However, isolated populations, such as Native Americans, have substantially reduced haplotype diversity. Thus, the power of discrimination is rather limited for casework and the effects of population substructure must be considered for some population groups. This study examined 12 additional highly polymorphic Y STR loci contained within two multiplexes to determine if these 12 Y STRs or a subset of the loci could obtain higher discrimination power. Four sample populations in the United States were examined: Native American (Alaskans), African American, Caucasian, and southwestern Hispanics. The following Y STRs were typed: DYS481, DYS576, DYS549, DYS643, DYS533, DYS570, DYS449, DYS460, DYS505, DYS522, DYS612, and DYS627. In addition, these samples were typed for the 15 autosomal STR loci using the PowerPlex® 16 HS system (Promega Corporation, Madison, WI). The data show that these additional Y STR loci are among the more discriminating Y STRs used in this study and are good candidates for increasing haplotype diversity. Population parameters, statistical applications, and novel null alleles and duplications will be presented. In addition, potential kit configurations will be discussed.