B157 Genetic and Population Characterization of the 17 Y STR YFiler Loci in Three Texas Population Groups

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This presentation will present population data and characteristics of the multiplex Y-Filer kit so that the forensic scientist can properly interpret Y-STR data. The approaches for interpretation of forensic Y STR haplotype evidence will be presented.

This presentation will impact the forensic science community by enabling scientists to properly evaluate and assess Y STR DNA evidence.

The Y-chromosome STR genetic markers can be useful for analyzing samples derived from violent crime because these targets are on the male-specific portion of the human genome. Additionally, these genetic markers can assist in resolving paternal lineage issues. The AmpFℓSTR®Yfiler™ Kit (Applied Biosystems) is particularly useful for such analyses, because it contains the reagents necessary for typing 17 Y STR loci in a single multiplex analysis. The loci are: DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385 (note that this marker represents two loci), DYS438, DYS439, DYS437, DYS448, DYS456, DYS458, DYS635 and Y GATA H4. In order to place proper significance regarding a 17locus Y STR haplotype obtained from a forensic specimen that matches a profile from a suspect or victim (or cannot be excluded as arising from a biological relative), population data sets are employed. These sample populations allow for: (1) estimating upper bounds on the frequency of an observed Y STR haplotype, (2) assessing the impact of population substructure on estimating the rarity of a haplotype, and (3) identifying any analytical characteristics of the genetic loci and multiplex systems that should be considered when effecting an interpretation. In this study approximately 3000 unrelated males residing in Texas, parsed out over three populations (African American, Caucasian, and Hispanic) were typed. This is the largest United States regional population analysis performed to date. Most of the time only one allele was observed per locus, except for the DYS385 marker where typically one or two alleles were observed. There were a few instances where multiple alleles per locus were observed. The DYS19, DYS439, DYS389I, and DYS385 loci exhibited the majority of multi-allele loci. In a very few samples there appeared to be a null allele at the DYS448 locus which occurred concomitantly with the observance of two alleles or an allele peak with increased height at the DYS437 locus. These samples are being sequenced to determine whether this observation is due to a deletion within the region where the DYS448 locus primers reside. All three population samples were highly polymorphic for the 17 Y-STR haplotypes. The haplotype diversity was greater than 99.9% for each population group. The genetic variance component analysis was conducted using the AMOVA routine of Arlequin 2.0. The Fst value across all three populations is exceedingly small (<0.001), such that given the population size of the databases, accommodating substructure will have little or no affect in estimating an upper bound frequency of Y STR haplotypes when a complete 17 locus profile is obtained. Y-STR mutations have important implications for parentage testing and for establishing male lineage in forensic applications. The population samples have matching father/son haplotypes confirmed by paternity testing (with high likelihood ratios). The average rate of mutation is consistent with other studies (i.e., the range of 10^{-3}/locus /generation). While DNA profiles based on Y-STRs cannot achieve individualization because they are paternally transmitted and non-recombining, large datasets enable better exploitation of the power of an analysis. These analyses provide guidance for an effective approach for estimating the rarity of a Y STR haplotype, and the approach will be discussed.

Y STRs, Multiplex, Population

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