



### **B43 Specific Haplotypes and Population Analysis of 17 Y-Chromosome STR Loci in Taiwan**

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After attending this presentation, attendees will understand the population frequencies (genetic polymorphism) and mutation of 17 Y-chromosome STR loci and specific haplotypes in the population in Taiwan.

This presentation will impact the forensic community and/or humanity by demonstrating that some specific haplotypes are found in the population in Taiwan, this information is important for a precise estimation of the frequency of duplicated mutated Y-STR alleles in forensic practice.

Y-Chromosomal short tandem repeats (Y-STRs) have been increasingly used, during the past few years, in the study of human evolution and human identification in forensic casework. Many commercially available kits have been adapted for forensic applications. This study was designed to establish the haplotype database in the Taiwanese population for the set of 17 Y-STR loci adapted by the AmpFLSTR Yfiler™ PCR Amplification Kit (Applied Biosystems, Foster City, CA, USA).

Two-hundred-three unrelated male Taiwanese were analyzed for the haplotypes in these 17 Y-STRs loci (DYS19, *DYS385a/b*, *DYS389I/II*, *DYS390*, *DYS391*, *DYS392*, *DYS393*, *DYS438*, *DYS439*, *DYS437*, *DYS448*, *DYS456*, *DYS458*, *DYS635* (Y GATA C4), and Y GATA H4) using the kit purchased from Applied Biosystems. The haplotype frequencies in these loci, gene diversities and the power of discrimination for each Y-STR locus were estimated. Mutation rates of these 17 Y-STRs were also examined by comparing the genotypes obtained from the father/son sample pairs.

One-hundred-ninety-two haplotypes were observed in the 203 unrelated males. No Y-STR mutation was found in 38 father/son pairs. Gene diversity (GD) and discrimination power (DP) values observed in these loci ranged from 0.399 (*DYS 391*) to 0.956 (*DYS 385*) and 0.397 (*DYS 391*) to 0.952 (*DYS385*), respectively. The GD values of five loci (*DYS 389 II*, *DYS 458*, *DYS 635*, *DYS 392* and *DYS 448*) were higher than those reported in the study including three different populations in the U.S., *i.e.*, African-American (N = 333), Caucasian (N = 254) and Hispanic (N = 175). The GD values of *DYS 391* and Y GATA H4 were lower than that reported by this same U.S. study. These results indicated that the AmpFLSTR Yfiler™ PCR Amplification Kit has improved minimal haplotype diversity and DP of the Y-specific haplotypes in the population studied and is suitable for forensic DNA analysis in Taiwan.

Of particular interest was the observation of one sample with four peaks at the *DYS 385* locus (a duplicated locus). A pedigree showing triple alleles at the *DYS 385* locus without allelic discrepancy between the father and son has been reported in a study on Japanese population. Double peaks for a single locus were also reported from Bahia (Brazil) including *DYS 389II*, *DYS 437* and *DYS 439*, presumably because of a duplication event followed by a mutation. Thus, the presence of double, triple or four alleles at one locus does not always implicate multiple male profiles.

**Y-STRs, Gene Diversity, Discrimination Power**