

CHARACTERIZATION OF THE PHILIPPINE Y STR POPULATION INVOLVING 23 LOCI: POPULATION INFORMATION, MUTATION ANALYSIS AND FORENSIC ASSESSMENT

Sheila Marie T. Angustia, Aquilina P. Pia, Franklin L. de la Cruz, Ma. Carmen Ablan- Lagman and Lorna H. Santos

Philippine National Police

Y- chromosomal STR markers were found to be significant in analyzing forensic DNA samples due to its male specificity, high sensitivity and paternal mode of inheritance. They have shown to be advantageous in analyzing male DNA in mixtures containing high female DNA fraction and as supplementary markers for kinship testing. To be useful in the Philippine population, markers must first be assessed for its variability and mutability.

In this study, twenty-three Y- chromosomal STR markers (DYS19, DYS385a/b, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635, Y-GATA-H4 DYS481, DYS533, DYS549, DYS570, DYS576, and DYS643) were used in characterizing the Philippine male population. Biostatistical properties (allelic frequency, haplotype diversity, gene diversity and discrimination capacity) was gathered and evaluated in 629 unrelated Filipino males to determine the usefulness of twenty-three Y STR markers in routine forensic analysis. Analyzing the data reveals that additional markers renders an increase in haplotype diversity, discrimination capacity and resolving power which exemplifies the forensic usefulness of additional markers in discriminating haplotypes from the Philippine population. Also, additional markers have demonstrated its ability to resolve common haplotypes to unique profiles. Information gathered from this study was submitted to the YHRD database.

Mutability of Y STR markers impacts interpretation of DNA profiles gathered during examination. While conventional Y STR markers having low mutation rates are useful in establishing relatedness among male individuals, rapidly mutating Y STR markers (RM Y STR) characterized by high mutation rates can be used to differentiate male persons from the same paternal lineage. Therefore, markers need to be correctly classified to determine its suitability on the case being investigated. In this research, mutability studies were conducted to obtain accurate data on mutation rates by scanning Y STR mutation on the Philippine population. Using one hundred thirty-three father and son transmission from previously confirmed paternity related cases, a total of eleven mutational events have been recorded with three mutations observed at DYS635. Among the 3,059 meiotic transfers covered, the 11 recorded mutations can be translated to an average overall mutation rate of 3.59×10^{-3} for the Philippine population. In addition, DYS635 which possesses high mutation rate (2.26×10^{-2}) is recommended to be reclassified as rapidly mutating.