

MOLECULAR ANALYSIS OF THE TWO HYPERVARIABLE REGIONS OF MITOCHONDRIAL DNA IN TAIWANESE

Tsang-Ming Ko, Pei-Tzu Wu, Jeff Chia-Kai Hsu, Pi-Mei Hsu, Yi-Pei Chung

Department of Medical Genetics, National Taiwan University Hospital, Taipei, Taiwan



Mitochondrial DNA (Mt-DNA) is inherited maternally. Variations of the two hypervariable regions (HV1 and HV2) of Mt-DNA have been used in population genetics and forensic examinations. In this study, we examined HV1 and HV2 sequences from 310 unrelated Taiwanese (ethnic Chinese Hans). Both regions were selectively amplified with polymerase chain reaction (PCR). For HV1, a forward primer (nucleotide position 15977 to 15999) and a reverse primer (16400 to 16422) amplified a DNA fragment of 446 basepairs; while for HV2, a forward primer (nucleotide position 26-47) and a reverse primer (410-433) amplified a DNA fragment of 407 basepairs. The PCR products were used for fluorescence DNA sequencing. In samples with either a transition of thymine to cytosine at position 16189 or with an insertion of one to three cytosines at position 309 after which a frame-shift phenomenon was often observed, DNA sequencing of the reverse strand was performed. With HV1 and HV2 sequences combined, 303 different Mt-DNA types were found. Only 7 types were shared by two samples. The informative rate obtained by DNA sequencing was higher than that obtained by haplotype analysis with restriction fragment polymorphism or oligonucleotide hybridization. The high heterogeneity of the Mt-DNA HV regions indicates that direct sequencing of these two segments is highly useful in human identification, sibship determination and forensic examination.

